

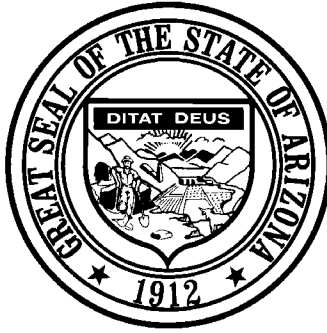
**ARIZONA DEPARTMENT OF HEALTH SERVICES
OFFICE OF WOMEN'S AND CHILDREN'S HEALTH**

GENETIC HEALTH CARE IN ARIZONA

A Plan for the 21st Century



April 12, 2000



**Jane Dee Hall, Governor
State of Arizona**

**Catherine R. Eden, Director
Arizona Department of Health Services**

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TABLE OF CONTENTS

Acknowledgments	ii
Abstract	iii
I. Executive Summary	I-1
II. Background and Purpose	II-1
III. Project Approach	III-1
IV. Needs Assessment Results	IV-1
A. Population at Risk	IV-1
B. Clinical Services	IV-14
C. Laboratory Services	V-28
D. Health Promotion and Preventive Health Services	IV-31
E. Social, Legal, and Ethical Issues	IV-38
F. Data Collection and Analysis	V- 47
V. Recommendations for Arizona Genetic Health Care Services	V-1
VI Appendices	
A. Organization Chart	A -1
B. Glossary	B -1
C. References and Resources	C -1
D. Program Desc, ADHS Office of Women's and Children's Health	D -1
E. Acronyms	E -1
F. Arizona Laws and Rules Regarding Genetic Services	F -1
G. Children's Rehabilitative Services Medically Eligible Conditions	G -1
H. Needs Assessment Research Questions	H -1
I. Genetics Plan Summary	I -1

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ABSTRACT

The ADHS Office of Women's and Children's Health and the Arizona State Genetics Services Advisory Committee (GSAC) collaborated in the planning and development of the Arizona Genetic Health Services Plan. The plan provides recommendations and a general plan to meet the challenge of assuring access to quality clinical genetic services for all families in the state of Arizona. The Arizona Genetic Health Services Plan focuses on key priorities that were identified through a systematic, yet dynamic self-assessment process. The plan considers the components of preconception, prenatal, children's and adult services. The following needs provide the foundation for the Arizona Genetic Services Plan:

1. The need for health care providers to be aware, informed, and educated regarding genetic services;
2. The need for community awareness and consumer information about birth defects, especially those that are preventable;
3. The need for individualized patient information and education regarding teratogen exposure, the benefits of preconception health planning, healthy behaviors during pregnancy, and availability and accessibility of genetic services;
4. The need to develop and support collaboration, cooperation, and service continuity among health care service networks, governmental agencies, and community agencies related to genetic services and the prevention of birth defects;
5. The need to improve data collection, integration and reporting capabilities related to: genetic risk assessment, genetic services, and incidence of genetic conditions;
6. The need to enhance Arizona's laboratory/service network for conducting certain types of genetic tests;
7. The need to assess the ongoing need for additional services, such as the Teratogen Information and Education Service; and
8. The need for Arizona to address "medical necessity" and insurance issues related to the availability and accessibility of genetic services;

The plan is intended to be used as a framework for guiding efforts to improve genetic health services for all Arizonans. The GSAC acts as the Steering Committee for the implementation of the plan. Standing committees of GSAC or other specialized committees are responsible for implementing the various components of the plan, and report findings and progress to the

Genetic Services Advisory Committee. Partnerships and collaborative efforts are undertaken with government agencies, public and private sector organizations, health care providers, educators, and consumers to achieve plan tasks and goals and standards.

The Arizona Department of Health Services, health care providers, and consumers have a critical role in the implementation of this plan.

ADHS's role is best suited to providing:

- Statewide coordination
- Public health education
- Leadership to obtain external funding
- Support of system linkages and integration
- Information system coordination
- Regulatory enforcement and accountability
- Program evaluation

Service providers should participate in:

- Standard setting for genetic service delivery
- Medical care delivery and quality assurance monitoring
- Research
- Ensuring availability and accessibility of genetic services
- Advocacy and support

Consumer roles include, but are not limited to:

- System information support
- Supporting information regarding consumer rights and responsibilities
- Practicing healthy behaviors (e.g., folate consumption in women of child-bearing age, and exercise and diet modifications for persons at risk of heart disease, diabetes, stroke, etc.)
- Seeking primary and supportive health care services
- Support and advocacy for persons with birth defects or genetic disorders
- Informing legislators about issues affecting the genetic service delivery system.

Health care agencies and human service organizations throughout Arizona are also critical to the success of this plan, for it is their collaborative partnerships that support the infrastructure of genetic service accessibility.

The needs assessment and technical plans presented in this document are to be used by Arizonans in formulating strategies for improving the quality of, and access to, genetic services. Other states are also encouraged to use this document as a resource in developing and implementing their own genetic service plans

I. Executive Summary

I. EXECUTIVE SUMMARY

In the last two to three decades, public health departments and medical genetics providers have developed a strong collaborative effort in establishing genetic programs. The primary purpose of this collaboration was intended to decrease morbidity and mortality related to birth defects and genetic disorders. As a result, several successful programs have been developed nationwide, such as newborn screening programs and outreach clinics. These services have resulted in increased access to genetic services for families who have, or who are at risk for, genetic disorders or birth defects. However, with the advent of health care reform, prevalence of managed health care systems, and changing role of public health, there may be barriers to the availability or accessibility of these genetic services. Advancements in technology over the last decade are resulting in a revolution of new DNA diagnostic tests. The new technology has not only allowed for the diagnosis of individuals affected with a specific single gene disorder, but is also promoting development of genetic tests to detect predispositions to common conditions such as cancer, cardiovascular disease, and mental disorders.

The document "Genetic Health Services in Arizona, A Planning Guide for the 21st Century" was developed by the Arizona Genetic Services Advisory Committee (GSAC). GSAC is a consortium of genetic health professionals, consumers, public health representatives, public and private sector organizations, health care providers, educators and other community stakeholders with an interest in the improvement of genetic health services for Arizonans. The project was funded by the Arizona Department of Health Services, Office of Women's and Children's Health. The planning guide provides ideas, potential strategies, and suggested recommendations for improving access to quality clinical genetic services for all Arizona families. The planning guide also provides recommendations for suggested roles for public health, genetic service providers, and consumers in the planning, implementation, and regulation of genetic services.

PROJECT APPROACH

The Arizona Genetic Health Services Planning Guide was developed through a dynamic, yet systematic process that included five major phases:

Phase I -- Planning and development of the needs assessment

A needs assessment established the foundation for suggested genetic services goals and recommendations. Six major steps were included in this planning phase:

1. Development of a working group or task force to oversee the process.

The Genetic Services Advisory Committee (GSAC) organized a task force to plan and implement the project. The GSAC Task Force was comprised of geneticists and genetic counselors from the University of Arizona, representatives from ADHS, private

practitioners, consumers, and consultants who were involved in program planning and project management with respect to Arizona's genetic services system.

2. *Development of the project framework and purpose.*

The GSAC met to outline critical features that the document would address. Types of questions considered during this process included, but were not limited to:

- What is a genetic services plan, and how would such a plan be used?
- What are the purposes and objectives of the plan or planning guide?
- What genetic issues need to be addressed for Arizona?
- What authority would a plan or planning guide have?
- What type of needs assessment is required, if any?
- What resources are needed to complete the needs assessment and the plan? (personnel, materials, expertise, funding)
- How can a plan be developed that will reflect the changing health care system?
- How can genetic issues be addressed?

3. *Review of current documentation and literature.*

The GSAC Task Force conducted a preliminary literature search and reviewed current documentation to identify existing guidelines and standards for genetic services. Information was requested and obtained from other states that had addressed genetic service issues from the state public health perspective.

4. *Documentation of issues to be addressed through the needs assessment.*

The GSAC developed an extensive list of "questions" regarding genetic issues in Arizona. These questions encompassed a wide variety of topics related to the organization, process, and implementation of genetic services. The questions were also utilized as part of the infrastructure for the development of the project plan.

5. *Definition of Goals and Objectives.*

Once the questions were categorized, and all the issues of interest had been documented by the committee, the issues of most concern were prioritized. Goals and objectives for the needs assessment were prepared from the prioritized issues.

6. *Development of needs assessment tools.*

As a precursor to the development of the needs assessment tools, current data sources

were identified to answer specific questions. Because this was the first time that a genetic needs assessment was performed in Arizona to address genetic services, the GSAC Task Force

recommended that the needs assessment address all areas of genetic need throughout the life cycle: preconception, prenatal, childhood, and adulthood.

In addition to existing data sources, several other key sources were used to develop needs assessment information: 1) discussions with consumers; 2) written surveys/questionnaires; and 3) telephone and personal interviews with providers, clients, and genetics professionals.

The Committee developed thirteen questionnaires for a variety of genetic service professionals, provider groups, and health care systems. In addition, a consumer forum was held to obtain information about relevant consumer issues. The questionnaires were completed using three methods; 1) written surveys completed by the respondent; 2) personal interviews, and 3) telephone interviews.

Phase II -- Implementation of the needs assessment

The assessment of needs was accomplished and coordinated through the GSAC. Committee members were assigned specific tasks. A lead coordinator was responsible for monitoring the status of the project, and progress toward completion of activities and group member assignments.

Phase III -- Compile and analyze needs assessment findings

Needs assessment findings were analyzed and summarized, and preliminary recommendations were developed. Results of the needs assessment were documented in a "self-assessment" format. For each of the major categories of attributes being studied, a best practice standard was developed, e.g., the "ideal situation" or "gold standard" relative to that topic. In developing potential goals and recommendations for genetic services, the group acknowledged that many of the standards would be difficult to achieve in the short term, and that some may not even be readily achievable in the long term. The idea was to create the ideal achievement standards for genetic service delivery, and a mechanism for measuring incremental progress toward attaining the ideal situation.

Phase IV -- Establish goals and recommendations, based on the needs assessment

Once the best practice standards were developed and documented for each attribute, the needs assessment results were outlined. Following an examination of the existing needs assessment information, task force members and consultants identified suggested action steps, and activities, that could move Arizona incrementally toward the best practice standards. Improvement indicators (performance indicators) were also developed for each standard. These indicators included a combination of structure, process, and outcome measures.

Phase V -- Write the plan

Creation of the needs assessment summary formed the basis for the preparation of recommendations and planning options.

Phase VI -- Implementation and Evaluation

This document is intended to be used as a planning document; a template to start discussion about how to prioritize and address genetic services in Arizona. Progress in developing and implementing goals and objectives will be reviewed. As plans of action are developed, they can be reviewed annually and revised as necessary to accommodate changing needs and priorities for Arizona.

THE ARIZONA GENETIC HEALTH SERVICES PLANNING GUIDE

The Arizona Genetic Health Services Planning Guide focuses on key priorities that were identified by the systematic self-assessment process. Components of preconception, prenatal, children and adult services were considered. There were several key themes arising from the needs assessment analysis that provide the foundation for the Arizona Genetic Services Planning Guide:

1. **The need for health care providers to be aware, informed, and educated regarding genetic services.** Health care providers include health care and social service providers at all levels: physicians, nurses, hospitals, insurance carriers, and adoption agencies. Providers need to realize that they collectively have tremendous potential for impacting the genetic health of Arizonans. Early intervention, age appropriate risk assessment, appropriate referral to genetic testing and diagnostic services, and early treatment, all make a difference.
2. **The need for community awareness and consumer information about birth defects, especially those that are preventable, and the availability of genetic services.** Community awareness activities should be culturally and linguistically appropriate, include the general public, and should also be targeted to all levels of schools and community organizations.
3. **The need for individualized patient information and education** regarding teratogen exposure, the benefits of preconception health planning, healthy behaviors during pregnancy, and availability and accessibility of genetic services.
4. **The need to develop and support collaboration, cooperation, and service continuity** among health care service networks, governmental agencies, and community agencies related to genetic services and the prevention of birth defects.

5. **The need to improve data collection, integration and reporting capabilities** related to: genetic risk assessment, genetic services, and incidence of genetic conditions.
6. **The need to enhance Arizona's laboratory/service network** for conducting certain types of genetic tests.
7. **The need to refine components of the needs assessment** to assess the ongoing need for additional services, such as the Teratogen Information and Education Service.
8. **The need for Arizona to address "medical necessity" and insurance issues related to the availability and accessibility of genetic services.** Preconception and genetic counseling, genetic evaluation, and multi-specialty, interdisciplinary adult clinics are usually not covered, because they are not considered medically necessary.

GENETIC HEALTH SERVICES PLANNING GUIDE ORGANIZATION

Recommendations for genetic health services in Arizona are derived from sixteen "gold standards". The gold standards were developed as high level "ideals" for a statewide genetics program. Some of the standards may be more difficult to achieve than others, but incremental progress for any gold standard would greatly improve the quality of the program. The standards were based on the prioritization of the key needs assessment findings.

These general recommendations are intended to be used to guide genetic service planning efforts in Arizona. The document is intended to be used as a stepping stone for further discussion, dialogue, and strategic planning for genetic services. We hope that there will be development of additional, specific projects and priorities, and ongoing evaluation of plans and processes related to genetic services.

ARIZONA GENETIC HEALTH SERVICES PLANNING GUIDE SUMMARY

The following genetic health services planning guide incorporates needs assessment findings and major recommendations for addressing those needs. This summary forms the foundation for more detailed plans that could be developed to improve genetic health services in Arizona.

ARIZONA NEEDS:

1. FEWER CHILDREN BORN WITH PREVENTABLE BIRTH DEFECTS

- **All women of childbearing age to be aware and informed about preventable birth defects, teratogens, and maternal illnesses that can affect pregnancy**
- **All women of childbearing age to take a daily folic acid supplement**
- **More people avoiding unhealthy behaviors and teratogen exposure**

Babies in Arizona are born with many different types of birth defects. For a number of these disorders, such as neural tube defects and fetal alcohol syndrome, there are ways to prevent or reduce the occurrence of these disorders in the newborn population. Arizona can continue to focus resources on preventing certain types of birth defects through early intervention, education, and anticipatory guidance. It is also possible to mobilize resources toward minimizing untoward effects and sequelae of many “non-preventable” genetic disorders. Primary prevention of disease in children can be accomplished through avoidance of behaviors or teratogens that put individuals at risk for producing offspring with birth defects. In conjunction with primary prevention efforts, ADHS will address secondary prevention through the development of systems and support for early identification, treatment, and prevention of complications from non-preventable genetic disorders. Tertiary prevention is aimed at minimizing the negative impact and effects of a genetic disorder to the child and family (e.g., early intervention programs for children with developmental delays) (Khoury, et al 1996).

ARIZONA COULD MEET THIS NEED BY:

- *Developing public/community-based training and education programs for providers in the detection of risks and methods of reducing preventable birth defects.*
- *Promoting prevention of fetal alcohol syndrome (FAS) and alcohol related birth defects through education and prevention programs.*

Promoting folic acid supplements for all women of childbearing age.

- Summarizing available research and information on preconception folic acid supplements and intake.
- Developing and implementing a Children's Information Center survey on folate and vitamin supplements.
- Reviewing Centers for Disease Control and Prevention (CDC) written guidelines for folic acid supplementation.
- Facilitating the development of clinical practice protocols for folic acid supplementation.
- Facilitating the development of clinical practice protocols for folic acid supplementation.
- Informing and educating the public about preventable birth defects.
- Expanding and enhancing Teratogen Information and Education Service (TIES).
- Working with the provider community, county health departments and consumers to identify and promote community-based, culturally appropriate information about teratogens.
- Promoting the dissemination of printed information in providers' offices.
- Promoting the use of public service announcements when appropriate.
- Working with providers, AHCCCS health plans, home visiting programs, and others, to target promotional efforts.
- Promoting collaboration with other resources and teams involved in health education and preventive care.

ARIZONA NEEDS:

2. PERSONS TO HAVE APPROPRIATE GENETIC RISK ASSESSMENT AND INTERVENTION FOR THE PREVENTION OF BIRTH DEFECTS AND GENETIC DISORDERS

- **Timely and appropriate referrals for genetic testing and services**
- **All pregnant women to receive a prenatal risk assessment for genetic disorders/birth defects and teratogen exposure**
- **Women of childbearing age and their partners to be offered screening for genetic disorders/birth defects**
- **Genetic risk assessment to be conducted in reproductive health settings and access to genetic information to be limited.**
- **All pregnant women (who are at or over the age of 35 at delivery) to be offered prenatal diagnosis for the detection of chromosomal abnormalities**

In Arizona, many women and their partners do not have access to preconception genetic screening services. Furthermore, many insurers do not cover preconception genetic testing services, because they are not considered to be “medically necessary”. Genetic testing and screening services may be more commonly considered by the provider community to have greater value after conception has occurred, or after the woman has already entered prenatal care. Prenatal care may not be started until 8 to 12 weeks into the pregnancy, which precludes the opportunity for education about the importance of prenatal vitamins with folic acid, and the reduction of teratogen exposure early in pregnancy.

Arizona could do more to promote use of preconception and prenatal risk assessments that address genetic disorders. It is thought that the most opportune time for prevention is in the preconception period when a woman has the opportunity to alter and adjust behaviors and practices, in order to minimize teratogen exposure and other threats to early embryonic development. Clinics and health plans which provide reproductive health, well-woman care and preventive services have the opportunity to access women of childbearing age prior to conception. It is possible to conduct age appropriate risk assessments that have a genetic screening component, as well as provide screening information about teratogen usage or exposure.

Many managed care plans in Arizona are developing, or have developed, health screening and risk assessment components that focus on preventive screening services for women of childbearing age, and adult health. The health screening and risk assessment should consider ethnicity, age, and family history. These areas can be more clearly identified, coordinated, and supported throughout the state, so that genetic risk screening can be a more consistent practice among all providers. The ultimate goal of this process is to develop a framework of standardized guidelines for providers to utilize in conducting broad-based genetic risk assessment activities.

ARIZONA COULD MEET THIS NEED BY:

- *Promoting early identification of the need for genetic testing and services through provider education, and community awareness.*
- *Surveying providers to determine risk assessment tools currently in use.*
- *Assessing utilization of genetic screening and testing services in reproductive health/family planning networks.*
- *Collaborating with health insurers regarding incidence of prenatal genetic risk assessment, and obtaining information about ongoing or completed studies regarding content of prenatal and preconception risk assessment activities.*
- *Assembling practice guidelines from other states regarding preconception, prenatal, and infertility testing.*
- *Defining "risk assessment", and the minimum elements to be included in a genetic risk assessment.*
- *Developing a plan to promote inclusion of a statewide genetic screening component into aspects of well woman*
- *care and preventive health services for women, children, and families.*
- *Identifying existing programs and agencies for collaborative links.*
- *Developing and strengthening collaborative links with agencies and programs that are monitoring early entry*
- *into prenatal care.*
- *Developing a plan with other agencies and cytogenetic testing providers to assess actual utilization of*
- *prenatal chromosomal diagnosis in women 35 years of age and older (who were offered testing).*
- *Monitoring the Arizona network for genetic counseling services.*
- *Promoting appropriate genetic risk assessment activities.*

ARIZONA NEEDS:**3. ALL INDIVIDUALS WITH A FAMILY HISTORY OF A GENETIC DISORDER TO BE OFFERED GENETIC COUNSELING AND EVALUATION**

We know there are many common adult diseases, such as cancer, diabetes, and cardiovascular disease, that have a genetic component. Although the cause of these chronic and potentially life threatening illnesses is multifactorial, many other diseases with a more direct genetic link have similar potential for devastating effects, such as cystic fibrosis, Huntington disease, or certain metabolic diseases. Very little is currently known about the numbers of persons in Arizona that are referred for genetic counseling and evaluation because they have a family history of a genetic disorder.

ARIZONA COULD MEET THIS NEED BY:

- *Promoting insurance company awareness of the health benefits that can accrue to their members by promoting preventive genetic services.*
- *Developing a training plan for Arizona health care providers in genetic services and referrals.*
- *Convening a committee or task force to develop additional information and data about the incidence of persons in Arizona who are offered genetic counseling and evaluation due to a family history of a genetic disorder.*

ARIZONA NEEDS:**4. GENETIC SPECIALISTS THAT ARE AVAILABLE, ACCESSIBLE AND GEOGRAPHICALLY APPROPRIATE TO THE AT RISK POPULATION**

Many of Arizona's rural counties and communities are geographically isolated from many types of health care services. Genetics providers do conduct clinics in a variety of rural and urban communities on a routine basis; however, the no-show rates for appointments are high in certain areas. More work needs to be done to identify strategies for improving availability and accessibility to care and services for all who need genetic counseling, evaluation, or testing services.

ARIZONA COULD MEET THIS NEED BY:

- *Involving community and tribal leaders in designing education and information programs that are community based, culturally appropriate, and accessible to the rural and tribal communities.*
- *Enhancing the genetic services network, and improving availability and accessibility of services.*
- *Continuing to strengthen links between geneticists and other health care providers, and incorporating other related agencies and teams to the network.*
- *Using the task force to identify options for improving network availability and accessibility.*
- *Continuing promotion of genetics consultation contracts through ADHS.*
- *Working with the genetics community, providers, consumer groups, and CRS to develop information for Arizona health care providers and consumers on the availability of genetic clinics, and the names and addresses of all genetic service providers in Arizona.*
- *Developing a plan to refine and clarify the availability and accessibility standards for genetic services in Arizona.*
- *Developing strategies in collaboration with consumers for reducing the number of "no-shows" .*

ARIZONA NEEDS:

5. GENETIC DIAGNOSTIC PROCEDURES AND GENETIC TESTS TO BE PERFORMED FOLLOWING COMPREHENSIVE GENETIC EVALUATION BY APPROPRIATELY TRAINED SPECIALISTS.

6.

Genetic diagnostic and testing procedures should be conducted only after a comprehensive genetic evaluation. These tests should be performed only by appropriately trained specialists who have met educational and/or training requirements in the technical, diagnostic, and interpretive aspects of the procedures.

ARIZONA COULD MEET THIS NEED BY:

- *Preparing a document outlining what consumers should look for in seeking genetic evaluation, genetic testing services, including qualifications of genetic professionals.*
- *Preparing and disseminating genetic testing resource lists to all health care providers.*

ARIZONA NEEDS:

6. HEALTH CARE PROVIDERS TO BE KNOWLEDGEABLE AND INFORMED ABOUT GENETIC RISKS, SERVICES AND RESOURCES

- **All providers to be knowledgeable about laboratory resources and specimen submission procedures**
- **All providers to be informed about selected genetic risks**
- **All providers to be aware and informed of how to access genetic services.**
- **Health and social service training programs to include a genetics component**

ARIZONA COULD MEET THIS NEED BY:

- *Improving laboratory submission for newborn screening and referral for other genetic tests.*
- *Collecting data regarding the number of unacceptable specimens currently submitted.*
- *Preparing a genetic test submission manual for providers and laboratory professionals containing laboratory testing requirements for common genetic tests and newborn screens.*
- *Developing a training plan for educating health care providers regarding specimen submittal, and training providers in the proper submission of specimens.*
- *Developing and/or enhancing ongoing reporting or data collection mechanism for tracking unacceptable specimens.*
- *Promoting early identification of need for genetic testing and services through provider education of genetic risks, and community awareness.*
- **Developing and strengthening collaboration and links with programs and agencies.**
 - *Collecting information about how referral processes to genetic services are initiated.*

- Identifying continuing education and inservice opportunities for physician providers.
- Documenting a plan for informing providers how to access genetic services.
- Developing professional education programs for preconception, prenatal, children, adult issues.
- Educating managed care medical directors on the need for primary care providers to offer their patients genetic diagnostic services.
- Educating providers about genetic risks and the referral process.
- Collaborating with consumer groups and parent/child advocacy organizations to educate providers about the availability of family support organizations.
 - Surveying health service, social service, and medical education/training programs in Arizona to obtain information about genetics curriculum content.
 - Promoting inclusion of enhanced genetics component in health services/medical education curricula.
 - Linking with others to identify opportunities for collaboration.
 - Developing a plan (with educational institutions) to target and improve areas in need of additional coverage in curricula.

ARIZONA NEEDS:

7. GENETICS LABORATORY SERVICES TO BE OF HIGH QUALITY.

- **Genetics laboratory services that are available, accessible, and timely**
- **Genetics laboratories that have appropriate quality control audits and/or accreditation**
- **Laboratory directors to be appropriately certified**

ARIZONA COULD MEET THIS NEED BY:

- *Improving the availability and accessibility of laboratory services.*
 - Convening a task force or subcommittee to explore options for development and support of additional diagnostic testing capability for genetic services in Arizona.
 - Identifying and documenting existing networks of genetics laboratory providers and facilities used by Arizona health care providers and organizations, and making the list available to the provider community through the education and orientation process.
 - Evaluating the current system, including specimen transportation, testing time, cost, protocol, interpretation of results, and other issues as identified.
 - Producing an evaluation report to the Genetic Services Advisory Committee regarding laboratory services in Arizona.
- *Assuring a mechanism for access to genetics laboratory testing when indicated, to ensure payment for genetics laboratory testing is available to all who require it.*
 - Convening a working group of the GSAC to explore options for payment, including clinicians, finance, managed care organization representatives, and AHCCCS.
- *Promoting appropriate accreditation for laboratory services that provide genetic screening and testing services for Arizona clients.*
 - Developing network and links with laboratory regulation agencies for genetics lab testing.

- Researching current requirements for laboratory certification in Arizona, and determining availability of other special certification/requirements for laboratories that handle metabolic or genetic testing specimens.
- Promoting the use of standards of practice by cytogeneticists and consultants to laboratories that provide genetic testing services through the inclusion of national practice standard requirements in existing state contracting systems (e.g. AHCCCS).

ARIZONA NEEDS:

8. INCREASED AWARENESS OF GENETIC ISSUES AND GENETIC SERVICES
ARIZONA COULD MEET THIS NEED BY:

- *Collaborating with providers, insurance companies, and consumers to document a plan for informing consumers on how to access genetic services.*
- Gathering information from insurance companies regarding the availability of internal genetics education programs.
- Gathering information from insurance companies and the provider community about how referral processes to genetic services are initiated for preconception, prenatal, child, and adult genetic services.
- Conducting a focus group for input regarding genetic service needs.
- Reviewing currently available consumer information.
- Developing a statewide genetic services awareness program with input from consumers and providers.
- *Developing public service announcements to increase awareness of preventable genetic conditions and availability of genetic services.*
- *Implementing a statewide genetic services awareness program.*
- *Seeking support for the project through the March of Dimes, Federal grants, or other funding sources.*

ARIZONA NEEDS:

- 9. ALL INDIVIDUALS TO BE INFORMED OF, AND THOROUGHLY COUNSELED AS TO THE BENEFITS, RISKS, AND LIMITATIONS OF GENETIC TESTING, DIAGNOSIS AND TREATMENT, AND THE POSSIBLE CONSEQUENCES OF GENETIC TESTING.**

ARIZONA COULD MEET THIS NEED BY:

- *Providing complete, accurate and clear consent information and other information for genetic testing.*
 - Conducting consumer, ADHS, and provider focus groups who would make recommendations and suggestions regarding informed consent for genetic testing.
- *Developing and promoting genetic education programs for providers that incorporate some instruction in genetic counseling.*
 - Reviewing currently available information during focus group meetings.
 - Developing educational materials to facilitate the provision of this instruction.
 - Developing and implementing multi-media training sessions and/or workshops to include opportunities for interactive practice in counseling about genetic services and testing.
- *Providing educational materials to providers/counselors regarding genetic testing and treatment options.*
- *Developing appropriate information and “minimum standards regarding benefits, risks, and options for genetic testing”, to be shared with clients, parents, and providers.*
- *Reviewing American Medical Association, American College of Obstetrics and Gynecology (ACOG), American Academy of Pediatrics, Alliance of Genetic Support Groups, American College of Medical Genetics, National Society of Genetic Counselors, and other guidelines for informed consent, and patient rights and responsibilities.*

- *Reviewing existing statements, and developing a statement of client rights and responsibilities related to genetic testing services.*
- *Providing client rights and responsibilities materials to providers and consumers.*

ARIZONA NEEDS:

10. ALL PERSONS TO BE PROTECTED FROM DISCRIMINATION RESULTING FROM GENETIC TESTING, SERVICES, OR DISORDERS. ARIZONA COULD MEET THIS NEED BY:

- *Educating the legislative community about genetic services, including preparing educational information in response to requests from various governmental agencies or legislative bodies.*
 - *Assisting in the development of appropriate educational materials for the legislature regarding newborn screening, and genetic lab testing.*
 - *Recommending improvements in legislation when needed.*
- *Promoting community integration of newly developed or approved legislation.*
- *Monitoring the implementation of new legislation.*
- *Developing systems and processes to ensure that genetic information is not used to make health care and other insurance prohibitively expensive.*
- *Prohibiting job discrimination based upon results of genetic testing, or the presence of a genetic disorder.*

ARIZONA NEEDS:

11. IMPROVED DATA COLLECTION AND ANALYSIS REGARDING BIRTH DEFECTS, GENETIC DISEASES, GENETIC SERVICES AND HEALTH OUTCOMES

ARIZONA COULD MEET THIS NEED BY:

- *Utilizing a working group of the Genetic Services Advisory Committee(GSAC) to conduct an in-depth analysis of genetic services data reporting requirements.*
- *Developing a core set of questions and issues regarding genetic services data, such as information regarding genetic risk assessment, genetic education, and utilization reporting.*
- *Studying genetics services data and reporting requirements from all systems: AHCCCS, state and private adoption agencies, Newborn Screening Program, CRS, specialty providers, and commercial insurers.*
- *Determining parameters, limitations, and restrictions for access to genetic services data and information.*
- *Creating a minimum data set of genetic services information, including a data dictionary (outlines definitions for data elements).*

ARIZONA NEEDS:

12. PERSONS TO HAVE ACCESS TO SINGLE SITE, MULTI-SPECIALTY, INTERDISCIPLINARY TEAMS WHEN APPROPRIATE FOR THE CARE AND TREATMENT OF IDENTIFIED GENETIC DISEASES.

Some children have access to multi-specialty, interdisciplinary care for several types of genetic disorders through the CRS/OCSHCN Program. Multi-specialty, interdisciplinary care for children with special health care needs considers the needs of the whole child and family, and provides continuity and consistency in the treatment of children with multiple medical or social needs. This care delivery system could also be evaluated for adult genetic services.

ARIZONA COULD MEET THIS NEED BY:

- *Obtaining more information about adult and pediatric multispecialty, interdisciplinary clinics.*
- *Developing a task force to examine multispecialty, interdisciplinary care options for genetic disorders.*
- *Identifying barriers for adults and children to access multispecialty, interdisciplinary care and services for genetic disorders (e.g., geography, personnel, cost, space and transportation).*
- *Developing and implementing a plan to improve multispecialty, interdisciplinary services and clinics for preconception, prenatal, children and adult services.*
- *Exploring options with CRS for expanding medical eligibility and funding for CRS services, by diagnosis first, then expanding for age after diagnosis.*

ARIZONA NEEDS:

13. ALL ADOPTIVE CHILDREN TO HAVE ACCESS TO GENETIC INFORMATION ABOUT THEIR BIOLOGIC FAMILY

- **All birth parents placing a child for adoption to be offered genetic screening****ARIZONA COULD MEET THIS NEED BY:**
 - *Reviewing current legal and legislative requirements for genetic information gathering for adoptions (public and private).*
 - *Reviewing national standards or guidelines from other states regarding collection of genetic information prior to adoption.*
 - *Identifying possible resources that could be used to assist birth parents to provide complete genetic information (e.g., questionnaire, family history taken).*
 - *Developing and implementing an educational program with adoption agencies regarding the importance of genetic information.*
 - *Working with state and private adoption agencies to implement options for improving non-identifying data collection of genetic information (family history or actual results of genetic testing and diagnosis) from birth parents to be provided to adoptive parents.*
 - *Evaluating if birth parents are supplying genetic information and if it is available to the adoptive child, and/or adoptive parents.*

ARIZONA NEEDS:

14. COMPLETE GENETIC BACKGROUND INFORMATION TO BE COLLECTED ABOUT EGG AND SPERM DONORS.

ARIZONA COULD MEET THIS NEED BY:

- *Reviewing current legal and legislative requirements for genetic information gathering for egg and sperm donation.*
- *Reviewing national standards or guidelines from other states regarding collection of genetic information prior to egg or sperm donation.*
- *Identifying possible resources that could be used to assist donors in providing complete information (e.g. questionnaire, family history taken), that would be available to egg and sperm recipients.*
- *Working with state and private adoption agencies, infertility clinics, and/or assisted reproduction facilities to investigate options for improving data collection of genetic information of birth parents.*
- *Developing and implementing an educational program, including a conference for infertility/OB providers, sperm bank staff.*
- *Implementing donor/carrier testing for common genetic disorders when available and provide appropriate follow-up.*
- *Evaluating if donors provide information and receive carrier testing and follow-up information.*
- *Evaluating surrogate mothers for metabolic disorders which might affect the fetal environment.*

ARIZONA NEEDS:

15. ALL PERSONS TO EXERCISE FREE CHOICE WITH RESPECT TO GENETIC TESTING OR TREATMENT

This “gold standard” was also identified as a core value of the Arizona’s genetic health care delivery system and plan.

ARIZONA COULD INCORPORATE THIS CORE VALUE BY:

- *Ensuring that use of genetic testing services is voluntary.*
 - Conducting surveys of providers who perform genetic tests, to determine whether genetic testing services are voluntary.
 - Assessing processes, procedures, and documentation produced and provided during pretest genetic counseling.
- *Investigating processes and procedures for providing post-procedure interpretation and counseling and guidelines for disclosure of test results and information.*
- *Promoting use of non-directive counseling.*
- *Developing an informed consent checklist (using input and review from geneticists and consumers) for*
- *counselors and providers to use in discussing genetics testing, treatment, and options with clients and families.*
- *Including genetics education themes in curricula, health services education, medical continuing*
- *education, consumer/prenatal education, provider training, patient Bill of Rights, and information about*
- *procedures to lay health workers.*
- *Incorporating information about genetic counseling, testing and treatment into training at all levels.*
- *Gathering policy statements related to presymptomatic genetic testing in children.*

ARIZONA NEEDS:

16. GENETIC SERVICES TO BE FAMILY CENTERED, CULTURALLY COMPETENT, INTEGRATED, GEOGRAPHICALLY APPROPRIATE, AND FOCUSED ON THE MAINTENANCE OF HEALTH

This “gold standard” was also identified as a core value of Arizona’s genetic health care delivery system and genetic health services plan.

ARIZONA COULD INCORPORATE THIS CORE VALUE BY:

- *Establishing a multidisciplinary task force/subcommittee to identify strategies for improving coordination of care and services for all persons who need or receive genetic services.*
- *Investigating current case management networks existing in Arizona for care and treatment of persons affected with genetic disorders.*
- *Enhancing communication and referral networks between rural primary care providers and urban based geneticists, multidisciplinary treatment specialists and insurance payers.*
- *Exploring the possibility of increasing access to information for the public and consumers, through technology such as Medline, Internet, or the ADHS Websites.*
- *Continuing to strengthen links with other health department teams, home visiting programs, family or consumer support groups, and other agencies.*
- *Exploring opportunities to expand state funding through CRS/OCSHCN for genetic testing for families in cases where definitive diagnosis for a child requires that family members be tested.*
- *Working with providers and parent support groups to develop and implement a confidential satisfaction survey for persons who receive genetic screening or testing services.*
- *Using results of the satisfaction survey to develop a plan for improvement.*
- *Working with wellness and prevention programs to identify strategies for wellness promotion in persons with genetic conditions.*
- *Promoting expansion of the family advocate and support network for service coordination resources.*

- *Identifying and linking with current follow up and coordination programs, and encouraging new programs where there are gaps in service.*
- *Preparing a Spanish translation for all written information provided to the public regarding genetic services.*
- *Training all professionals, including medical providers, hospital personnel, and teachers, in cultural awareness and sensitivity issues surrounding genetic services.*
- *Promoting the availability of verbal translation services and service coordination to meet the needs of individuals seeking genetic services.*
- *Considering availability of translation services for tribal populations, and appropriateness of information in written or spoken languages.*
- *Promoting integration and coordination of genetic services with other aspects of primary care.*
- *Promoting prevention and health maintenance attributes of genetic services.*
- *Developing sensitivity training programs for providers that support holistic treatment of children and families, incorporating the principles of family centered care.*
- *Involving consumer advocacy groups, health plan/insurance member service representatives, and other family advocacy agencies in efforts.*
- *Involving support groups and consumer advocates in designing materials for the genetics program and the public.*
- *Developing documentation for consumers and the public that is user friendly and easy to read.*
- *Promoting education in all sectors of the population regarding the genetic contribution to both common and rare diseases*
- *Involving community and tribal leaders in designing education and information programs that are community-based, and accessible to the rural and tribal communities.*

SUMMARY OF PUBLIC HEALTH, PROVIDER, AND CONSUMER ROLES

The public health services, health care providers, and consumers have a critical role in the implementation of any plans or strategies to improve genetic health services.

The public health role is well-suited to providing:

- Statewide coordination
- Public health education
- Leadership to obtain external funding
- Support of system linkages and integration
- Information system coordination
- Regulatory enforcement and accountability
- Program evaluation

- Providers can consider participating in:
 - Standard setting for genetic service delivery
 - Medical care delivery and quality assurance monitoring
 - Research
 - Ensuring availability and accessibility of genetic services
 - Advocacy and support

- Consumer roles may include, but are not limited to:
 - System information support
 - Supporting information regarding consumer rights and responsibilities
 - Practicing healthy behaviors that promote prevention of birth defects, reduce risks of developing genetic disorders
 - Seeking primary and supportive health care services
 - Support and advocacy for persons with birth defects or genetic disorders
 - Informing legislators about issues affecting the genetic service delivery system.

Health care agencies and human service organizations throughout Arizona are also critical to the success of this plan, for it is by their partnership and collaboration that the infrastructure of support for genetic service accessibility is built.

The needs assessment and recommendations presented in this document are to be used by Arizonans in formulating, prioritizing, and implementing more specific strategies for improving the quality of, and access to, genetic services. Other states are also encouraged to use this document as a resource in developing and implementing their own genetic service plans.

II. Background and Purpose

II. BACKGROUND AND PURPOSE

The current wave of revolutionary breakthroughs in genetic diagnosis and technology has had a significant impact on the availability of testing for specific gene disorders. It is now possible to test people for specific gene disorders, as well as for predispositions to common, yet potentially devastating conditions, such as cancer, cardiovascular disease, and mental health disorders. With the advent of “high-tech” genetics, there are also limitations and constraints that society must address. Should this technology be available to everyone, regardless of their ability to pay? Who pays? With the concurrent struggles to manage rising health care costs, along with cost-cutting and resource conservation by insurance companies and managed care organizations, access to this genetic technology is threatened. The Arizona Department of Health Services (ADHS) has recognized a need to identify strategies and plans to support and enhance genetic services for Arizonans. What is the role of public health? What are the roles of providers and consumers? How can ADHS impact the public health and welfare in a positive manner, to improve the quality of life for Arizonans who need genetic services?

Over the last several years, these and other questions have been raised by ADHS and the genetics community regarding funding, availability and accessibility, and the future of genetic services in Arizona. These primary questions are:

1. Do all families throughout the state of Arizona have access to quality genetic services?
2. What are the barriers to genetic health care?
3. Is current funding administered through ADHS being distributed efficiently and effectively to maximize access to care and improve quality of services?
4. Is Arizona planning for the future of genetic services, considering the advancements in technology and the impact of managed care on the health care delivery system?
5. Are social, legal and ethical issues resulting from the accelerating development of DNA-based diagnostic tests being addressed appropriately both by public health and medical providers?
6. What is the role of public health services in the planning, implementation, and regulation of clinical genetic services? What are the roles of providers and consumers?

Arizona has a long term investment in the planning, funding and implementation of genetic services. In collaboration with the Section of Medical and Molecular Genetics, in the Department of Pediatrics at the University of Arizona Health Sciences Center and other health care providers, ADHS has been very successful in establishing genetic screening and clinical services throughout the state, such as OCSHCN/CRS (Children's Rehabilitative Services) clinics and Arizona's Newborn Screening Program.

A Genetic Services Advisory Committee was formed to identify and address issues related to Genetic Services in Arizona. The Office of Women's and Children's Health (OWCH) at the ADHS and the Arizona Genetic Services Advisory Committee (GSAC) decided to collaborate in the planning and development of the Arizona Genetic Health Services Plan. The GSAC is comprised of ADHS/OWCH personnel, University of Arizona geneticists and personnel, private genetics practitioners, consumers, and other health care professionals. The Committee's purpose is to provide direction for planning, monitoring and funding genetics activities for the state of Arizona. The GSAC is concerned about promoting and improving access to quality genetic health services for Arizonans.

A subcommittee of the GSAC (GSAC Task Force) was established and a genetic consultant was hired. The Task Force was comprised of genetic service providers from the public and private sector, ADHS personnel and consumers.

The goals of the Plan are:

- 1) to meet the challenge of assuring access to quality clinical genetic services for all families in the state of Arizona who have, or who are at risk for genetic disorders.
- 2) to determine the role of the public health, genetic service providers, and consumers in the planning, implementation and regulation of genetic services.

The long term goals of genetic service programs are: to increase access to genetic health services for individuals and families, promote quality clinical genetic services, and to decrease morbidity and mortality associated with genetic disorders. The Genetic Health Services Planning Guide includes suggested recommendations for strategic goals, and incorporates suggested action steps for achieving goals. Evaluation criteria are offered to measure the progress of implementation. The following chapter presents the project approach used by the Task Force to develop and document the needs assessment and the planning guide.

ADHS and the GSAC will use this document as a resource to prioritize and mobilize resources toward improving statewide genetic health services. Other states are encouraged to use this document as a reference in developing their own genetic health service plans.

III. Project Approach

III. PROJECT APPROACH

The recommendations in this document were developed through a dynamic, yet systematic process that included five major phases. Each is briefly described in the paragraphs that follow. This description may provide a framework or starting point for other states who are considering developing recommendations and plans for genetic services.

Phase 1 -- Planning and development of the needs assessment

This phase is considered to be the cornerstone of the entire project. Iterative planning and development of the needs assessment establishes the foundation for recommendations, strategies, and suggested actions. Careful attention to the entire planning process, its timing, resources, and objectives, is also paramount. The ADHS Office of Women's and Children's Health (OWCH) and the Genetic Services Advisory Committee incorporated six major steps into this planning phase:

1. Development of a working group or task force to oversee the process.

The Genetic Services Advisory Committee (GSAC) organized a task force to plan and implement the project. The GSAC Task Force was comprised of geneticists and genetic counselors from the University of Arizona, representatives from ADHS, private practitioners, consumers, and consultants who were involved in program planning and project management with respect to Arizona's genetic services system.

2. Development of the project framework and purpose.

The GSAC met to outline critical features that the genetic services plan would address. Types of questions considered during this process included, but were not limited to:

- What is a genetic services planning document, and how would it be used?
- What are the purposes and objectives of the plan/planning guide?
- What genetic issues need to be addressed for Arizona?
- What authority is required to implement a genetic services plan?
- What type of needs assessment is required, if any?
- What resources are needed to complete the needs assessment and the plan? (personnel, materials, expertise, funding)
- How can planning guide recommendations reflect the changing health care system?
- How can genetic issues be addressed?

3. Review current documentation.

The Task Force conducted a preliminary literature search and reviewed current documentation to identify existing guidelines and standards for genetic services. Information was requested and obtained from other states who had addressed genetic

service issues from the state public health perspective.

4. Documentation of issues to be addressed through the needs assessment.

The GSAC developed an extensive list of “questions” regarding genetic issues in Arizona. These questions encompassed a wide variety of topics related to the organization, process, and implementation of genetic services. Once developed, the questions were categorized across several parameters: A) Population at risk; B) Clinical Services; C) Laboratory Services; D) Health Promotion and Preventive Services; E) Social, Legal, and Ethical Issues; and F) Data Collection and Analysis. Consumer issues were also addressed throughout the analysis. See the Appendix for a detailed list of these questions. The GSAC also identified four major life cycle stages that were important to examine: 1) Preconception; 2) Pregnancy; 3) Childhood; and 4) Adulthood, as well the implications for population-focused public health.

5. Definition of Goals and Objectives.

Once the questions were categorized, and all the issues of interest had been documented by the committee, the issues of most concern were prioritized. Goals and objectives for the needs assessment were prepared from the prioritized issues.

6. Development of needs assessment tools.

As a precursor to the development of the needs assessment tools, current data sources were identified to answer specific questions. Because this was the first time that a genetic needs assessment would be performed in Arizona, the GSAC Task Force recommended that the needs assessment address all areas of genetic need throughout the life cycle: preconception, prenatal, childhood, and adulthood. Data sources used included:

- a. Descriptive documents
- b. Clinical genetics regional data
- c. Newborn screening data
- d. Arizona Vital Records
- e. Arizona Birth Defects Monitoring Program registries
- f. Arizona Cancer Registry
- g. Other clinical service data (including OCSHCN clinics, disability, etc.)
- h. Medicaid Health Plan information from Arizona Health Care Cost Containment System (AHCCCS).
- i. Mountain States Regional Genetic Services Network Database
- j. ADHS Vital Statistics
- k. Consumer forum results

In addition to existing data sources, several other key sources were used to develop needs assessment information: 1) discussions with consumers; 2) written surveys/questionnaires; and 3) telephone and personal interviews with providers, clients, and genetics professionals.

The Committee developed thirteen questionnaires for a variety of genetic service professionals, provider groups, and health care systems. In addition, a consumer forum was held to obtain information about relevant consumer issues. The questionnaires were completed using three methods; 1) written surveys completed by the respondent; 2) personal interviews, and 3) telephone interviews. The questionnaires included the following:

1. Adoption
2. Reproductive Service Questionnaire for OB-GYNs and Agencies
3. Reproductive Service Questionnaire for Infertility Clinics
4. Reproductive Service Questionnaire for Perinatologists
5. Cytogenetic Questionnaire for Laboratories
6. Maternal Serum Screening Questionnaire for Laboratories
7. DNA Questionnaire for Laboratories
8. Children's Rehabilitative Services (CRS)
9. Geneticist Questionnaire for Pediatric Services
10. Geneticist Questionnaire for Adult Services
11. Specialty Clinic Questionnaire (for non-CRS clinics)
12. Pediatric Services Questionnaire for physicians and agencies (AHCCCS Health Plans)
13. Prenatal Services Questionnaire for physicians and agencies (AHCCCS Health Plans)

Phase II -- Implementation of the needs assessment

The assessment of needs was accomplished and coordinated through the GSAC. Committee members were assigned specific tasks. A lead coordinator was responsible for monitoring the status of the project, and the progress toward completion of activities and group member assignments.

Phase III -- Compile and analyze needs assessment findings

Needs assessment findings were analyzed and summarized, and preliminary recommendations were developed. Results of the needs assessment were documented in a "self-assessment" format. For each of the major categories of attributes being studied, a best practice standard was developed, e.g., the "ideal situation" or "gold standard" relative to that topic.

Phase IV -- Establish goals and recommendations, based on the needs assessment

Once the best practice standards were developed and documented for each attribute, the needs assessment results were outlined. Following an examination of the existing needs assessment information, task force members and consultants identified recommendations, action steps, and activities, that could move Arizona incrementally toward the best practice standards. Improvement indicators (performance indicators) were also developed for each standard. These indicators included a combination of structure, process, and outcome measures.

Phase V -- Write the plan

Creation of the needs assessment summary formed the basis for the preparation of recommendations and planning options.

Phase VI -- Implementation and Evaluation

This planning document provides the template for the prioritization and implementation of activities to improve access to, and quality of, genetic services for Arizonans. This document is intended to be used as a planning document; a template to start discussion about how to prioritize and address genetic services in Arizona. Progress in developing and implementing goals and objectives will be reviewed. As plans of action are developed, they can be reviewed annually and revised as necessary to accommodate changing needs and priorities for Arizona.

III. Needs Assessment Results

IV. NEEDS ASSESSMENT RESULTS

This Chapter presents a summary of ADHS's genetic services needs assessment. As described in Chapter III, the needs assessment focused on an analysis of six attributes: a) population at risk; b) clinical services; c) laboratory services; d) health promotion and preventive services; e) social, legal, and ethical issues; and f) data collection and analysis. Consumer issues related to these attributes are also incorporated throughout the analysis. These attributes were assessed with consideration to the life cycle (i.e., preconception, prenatal, childhood and adult issues). It is important to note, however, that most of the issues identified through the needs assessment were applicable to more than one life cycle phase.

A. POPULATION AT RISK

ARIZONA HAS:

GEOGRAPHIC, ENVIRONMENTAL, ECONOMIC AND CULTURAL DIVERSITY

Geographic and Environmental Diversity

Arizona is a state with tremendous geographic and environmental diversity. Arizona is the sixth largest state in the United States, but ranks 24th in total population. Fifteen counties cover an area of over 114,000 square miles. The state includes 21 Indian reservations, with more than 20 tribes native to Arizona. The majority of the population (approximately 75%) are located in the major metropolitan areas of Phoenix and Tucson, two mid-size cities of Yuma and Flagstaff, and numerous smaller communities. The average population density is 31.9 persons per square mile and ranges from 3.3 persons per square mile in La Paz County to 224.8 persons per square mile in parts of Maricopa County. Six Arizona counties may be described as frontier areas, with a population density of less than 7.0 people per square mile. One of the greatest barriers to health care services in Arizona is the geographic settlement pattern. In remote communities, rural residents must travel great distances to use even primary care services.

Economic Diversity

Although several communities in Arizona boast an above average per capita income, the overall average per capita income in Arizona is less than the national average. In 1989, Arizona ranked 28th among 50 states, with a per capita disposable income of \$13,669, more than 8.5% below the national average of \$14,945. Based on the 1990 U.S. Census, approximately 15.7 percent of Arizona's population have incomes below the federal poverty level. More than 36 percent have incomes below 200% of federal poverty level. Poverty level incomes are more prevalent among Hispanic, African American, and Native American families. Child poverty in Arizona has increased significantly in Arizona over the last decade.

An assessment of the State of Arizona, conducted by the ADHS Office of Women's and Children's Health in 1996 identified significant health system needs in rural areas of Arizona (OWCH, 1996). Many women and children continue to live in poverty, and a disproportionate share of these uninsured or underinsured Arizonans live in rural and frontier areas. The combination of low population density and lack of health insurance options makes it difficult for some rural communities to support full-time physician practice and/or specialty services.

Although Arizona as a whole has an adequate supply of health care providers, facilities and equipment, the distribution of these providers is heavily skewed to the urban areas. This makes access to specialized health care services difficult for rural areas. It is not uncommon for people to have to travel more than 45 to 60 miles to see a doctor. In some cases, rural residents use facilities in Utah, California, Colorado or New Mexico because of the geographic isolation of their community to Arizona's urban areas.

Perhaps more than any other state, Arizona's health care system emphasizes managed care, both in the public and private sectors. The Arizona Health Care Cost Containment System (AHCCCS, pronounced "access") provides health care insurance for approximately 470,000 members. Persons are eligible for AHCCCS through Title XIX (Medicaid) or other state or county-funded medical assistance programs. Through a competitive bidding and negotiation process, health plans compete for prepaid capitated contracts to provide care to eligible individuals. Health plans, in turn, contract with health care providers to provide covered services. One out of five children in Arizona, 19 years or younger, is enrolled in AHCCCS.

TABLE IV.1 POPULATION AND POVERTY POPULATION BY RACE/ETHNIC GROUP, AZ, 1990

Race/Ethnic Group	% of Arizona Population	% of People with Income at or Below Federal Poverty Level	% Neonates in 1996*
White, non-Hispanic	71.1	8.5	47.0
Hispanic	18.8	20.8	39.9
Native American	5.6	43.1	7.0
African-American	3.0	25.2	4.1
Asian/Pacific Islander/Other	1.5	2.4	1.9

SOURCE: 1990 U.S. CENSUS

* VITAL STATISTICS, 1996

Cultural Diversity

Arizona's rich cultural diversity includes the strong influence of Hispanic and Native American populations. According to the 1990 U.S. Census, nearly 19 percent of Arizona's population is Hispanic, and 5.6% of the population is Native American. Nearly 40 percent of infants born in 1996 in Arizona were Hispanic. Arizona is one of four states that border Mexico. English, Spanish, and Navajo are the most common primary languages spoken by Arizona residents. Migrant and seasonal farm workers are a special minority population. Various agencies estimate that there are between 32,000 to 90,000 migrant farm workers and their family members, concentrated primarily in the counties of Pima, Pinal, Maricopa, Yuma and Cochise. Although the majority of these farm workers are U.S. citizens, many Mexican citizens also cross the border daily for employment.

Many persons in minority and ethnic cultural groups have experienced barriers to receiving necessary health care services. A 1992 study by Arizona State University's Morrison Institute for Public Policy found an unmet need for linguistically and culturally competent providers in rural areas. These barriers present themselves also in urban areas, particularly for those minorities living in low-income neighborhoods, such as South Phoenix in Maricopa County, which includes the largest number of African Americans in the state, and a large Hispanic population.

Arizona also has an established health care system for Native Americans. The Indian Health Service (IHS) administers federal funds to improve the health of Native Americans. The Bureau of Indian Affairs (BIA) administers some funds for social service and substance abuse programs. Additionally, many tribes manage federally funded health and social programs in their own communities. As a result, on-reservation services are available to people of Native American descent who are members of that reservation community (definitions vary among tribes). Eligibility for urban populations is more liberal. Native Americans living on reservations in Arizona who also qualify for AHCCCS may either choose an AHCCCS health plan or IHS as their managed care provider. Native American people often report that services and providers both on and off reservations need to be more sensitive to their cultural beliefs and practices, including identifying opportunities to integrate traditional and modern "Western" medicine to improve health.

Community attitudes about health care, education, lifestyle preferences, social structure, and religious preferences and customs all contribute to make Arizona a diverse, complex, and challenging health service delivery system network.

ARIZONA HAS:

WOMEN AT RISK FOR GIVING BIRTH TO A BABY WITH A BIRTH DEFECT:

- **ALL WOMEN WITH A 3-4% BACKGROUND RISK**
- **MORE OLDER WOMEN GIVING BIRTH**
- **WOMEN USING ALCOHOL DURING PREGNANCY**
- **WOMEN USING DRUGS AND OTHER TERATOGENS DURING PREGNANCY**

The reported incidence of birth defects in Arizona's children indicates that there are babies who are being born with preventable birth defects. There were 971,606 women in 1996 in Arizona who were between the ages of 15 and 44 years, or childbearing age, as defined by the Maternal and Child Health Bureau and the Center for Disease Control (1996 Vital Statistics). Approximately 7.7 percent of women in Arizona gave birth in 1996 for a total of 75,094 births. There were 487,956 women in Arizona between 35 and 49 years of age during 1996. Approximately 10.4 percent of the births in Arizona were to women age 35 and older.

The incidence of birth defects in Arizona's children indicates that there are babies born with many different types of genetic disorders, several of which are preventable, such as neural tube defects and fetal alcohol syndrome. Women who are at risk for giving birth to a baby with a birth defect fall into one of several risk categories, those with: 1) background risk for congenital anomalies, 2) increased maternal age, 3) alcohol use, and 4) other teratogen use, such as illegal or prescription drugs:

- 1) "Background risk" refers to the statistical, inherent (population based) risk for a child to be born with a birth defect. Background risk is not a preventable risk factor.
- 2) Advanced maternal age, considered to be 35 years of age or older during the pregnancy.
- 3) Alcohol use during pregnancy.
- 4) Women with other teratogen exposure during the pregnancy. Teratogens include prescription and recreational drugs and other chemicals or substances that may have a harmful effect on the developing fetus.

Arizona has women with a background risk for giving birth to infants with congenital anomalies.

Congenital anomalies are among the leading causes of fetal and infant mortality in the United States, as well as Arizona. Approximately 3 to 4% of all newborns have a significant birth defect noted at birth (Jones, 1996). According to Arizona Vital Statistics for 1996, the incidence of births reported with congenital anomalies increased from 1990 to 1995 in Arizona. In 1995, 1,245 instances of birth defects were reported (duplicated count). In 1996, there were 1,379 births with birth defects, or 1.8 percent of all births. In 1996 in Arizona, the overall incidence of birth defects ranged from 2.6 percent among American Indians, 2.6 percent among African-Americans, 2.1 percent among Hispanics, to 1.5 percent among white non-Hispanics. (VS, 1996)

Congenital anomalies are the second leading cause of death for children from birth to age one year, second only to prematurity in the neonatal period (birth through 27 days), and SIDS in the postneonatal period (28 days up to 1 year). For the year 1996, the Arizona Child Fatality Review Team reviewed a total of 879 deaths of children between the ages of birth through 17 years. Of these 879 deaths reviewed, 155 (17.6%) of the deaths were due to congenital anomalies, as follows:

Period	Number of deaths reviewed	% of Deaths Reviewed due to Congenital Anomalies	Rank as a leading category of death
Neonatal (birth-27 days)	318	30.2%	2
Postneonatal (27days to 1 year)	178	20.2%	2
1-4 year olds	112	8.6%	3
5-9 year olds	51	11.8%	4
10-14 year olds	82	4.9%	5
Total, all age groups, birth-17	879	17.6%	

ARIZONA CHILD FATALITY REVIEW TEAM, THIRD ANNUAL REPORT, 1996

Medical conditions as a cause of death were reviewed in the Child Fatality Review Program's 1996 analysis, and included 6 cases of death due to metabolic disorders, which are often genetic conditions. Of the 254 cases reviewed by the Child Fatality Review Team that were preventable, 5 of the deaths were caused by congenital anomalies.

Several of these genetic disorders are thought to be "multifactorial", produced through a combination of genetic predisposition, and other environmental factors. One example of a birth defect in this category is a neural tube defect, such as spina bifida. Although there is a genetic component to this defect, and the risk of developing neural tube defects can be greatly reduced by

folic acid/folate intake in women of childbearing age.

Although many birth defects may not be preventable, Arizona can continue to focus resources on the birth defects that are preventable, through early intervention, education, and anticipatory guidance. It is also possible to mobilize resources toward minimizing untoward effects and sequelae of many non-preventable disorders. Primary prevention can be accomplished through avoidance of behaviors or teratogens that put individuals at risk for producing birth defects. In conjunction with primary prevention efforts, ADHS can address secondary prevention through development of systems and support to early identify, treat, and prevent complications of non-preventable genetic disorders (e.g., treatment of PKU by dietary modification). Tertiary prevention is aimed at minimizing the negative impact and effects of a genetic disorder to the child and family (early intervention programs for children with developmental disabilities). (Khoury, 1996 AmJPH)

Arizona has an increasing number of older women giving birth.

Approximately 10.4% of Arizona births in 1996 were to women who were age 35 or older (VS, 1996). The following table presents maternal age in Arizona, by region and ethnicity.

TABLE IV.2 MATERNAL AGE BY REGION AND ETHNICITY, ARIZONA, 1996

Maternal age	TOTAL	AREA		ETHNICITY				
		URBAN	RURAL	WHITE	HISPANIC	BLACK	AM. INDIAN	ASIAN
< 15	216	185	31	40	132	10	28	3
15-17	4340	3598	742	1368	2308	220	401	36
18-19	6691	5451	1240	2625	3188	268	567	37
20-24	21026	17413	3623	9209	9115	753	1674	262
25-29	20110	17020	3090	11054	6827	527	1212	453
30-34	14879	12703	2176	9215	3963	365	909	404
35-39	6549	5494	1054	4212	1549	163	426	188
40-44	1219	976	243	741	296	30	106	42
45+	46	35	11	28	13	0	3	2
UNKNOW N	9	4	5	2	5	0	2	0
TOTAL	75094	62879	12215	38494	27396	2336	5328	1427

SOURCE: ARIZONA VITAL STATISTICS, 1996

Approximately one in four mothers in 1985 were at least 30 years old compared to about one in three in 1996. More women than ever are choosing to delay pregnancy until they are well into their thirties, and although the general health status of the women may be excellent, there are increased risks to the mother and the pregnancy. Maternal age of 35 or greater in the pregnancy is considered to be "advanced maternal age" and is associated with a higher incidence of birth defects such as Down syndrome and other chromosomal abnormalities. The American College

of Obstetrics and Gynecology (ACOG) recommends offering prenatal diagnosis to women who are age 35 and older, to screen for chromosomal disorders.

Arizona has women at risk for giving birth to babies with preventable birth defects due to teratogen exposure via alcohol or drugs.

Alcohol consumption during pregnancy is associated with fetal alcohol syndrome (FAS), which causes mental retardation, developmental delays, and behavioral problems in affected infants, children, and adolescents. The estimated incidence of FAS in the population is approximately one in one thousand births. During 1986 through 1990, the years for which data are available through the ABDMP, there were 89 cases of FAS reported (ADHS; 1990 Birth Defects Monitoring Program Report, Epidemiologic Report Series 1997:4). In these years there were 327,669 live births and fetal deaths in Arizona, for a rate of 0.27 FAS cases per 1,000. In 1995, when there were 72,883 live births and fetal deaths (ADHS Vital Statistics, 1995), approximately 20 cases of FAS would have been expected using the 1986-1990 rate. Since the ABDMP ascertains cases up to 1 year of age only, this number probably falls short of the actual number of cases born in 1995. The latter is likely to be an undercount, as many women may not be accurately reporting their drinking habits. In addition, the diagnosis of FAS or fetal alcohol effects (FAE) is difficult to make. No lab tests can confirm the diagnosis, which is made on a subjective assessment of physical appearances and characteristic features, as well as historical report of maternal alcohol ingestion during pregnancy.

Indian Health Service reports that nearly 85% of its referrals to genetic services were to rule out FAS. Despite the efforts of public health programs and the health care community, many women of childbearing age are continuing to need treatment for alcohol and substance problems. ADHS needs more information in this area to be able to better target its prevention strategies and action plans where they will have the most benefit.

Arizona also has pregnant women who are using teratogens other than alcohol, such as illegal street drugs (e.g., cocaine, methamphetamine, etc.), or prescription medications. Some drugs used during pregnancy may produce birth defects, by affecting fetal development or intrauterine growth. Prescription drugs such as anticoagulants (e.g., Coumadin), and cystic acne medications (e.g. Accutane) should be avoided during pregnancy. Use of drugs and medication should be reconsidered in light of a pregnancy, and use of either prescription or over-the-counter drugs should be minimized, especially use of multiple drugs with unknown interactions.

ARIZONA HAS:

PRENATAL PROVIDERS WHO CONDUCT RISK ASSESSMENT FOR TERATOGEN USE Most obstetrical care providers in Arizona conduct some type of prenatal risk assessment for pregnant women. Currently, Hollister and American College of Obstetrics and Gynecology (ACOG) risk assessment tools are most commonly used for this purpose. These risk assessment tools focus largely on drug, alcohol, and smoking habits. MSRGSN has a more comprehensive risk assessment tool for identification of teratogenic, ethnic, age, or other potential genetic risk factors. We do not know what percentage of providers use the tools routinely and consistently, and it is not known whether providers have developed additional assessments related specifically to prenatal genetic risk screening.

It is critical for women to access prenatal care as early in their pregnancies as possible. It is even better if women of childbearing age can plan for the possibility of pregnancy by developing healthy behaviors and lifestyle practices in the preconception period. It is thought that the most opportune time for prevention is in the preconception period when a woman has the opportunity to alter and adjust behaviors and practices to minimize teratogen exposure and other threats to early embryonic development. Clinics and health plans who provide reproductive health and well-woman care and preventive services have the opportunity to access women of childbearing age prior to conception, and if possible, conduct age appropriate risk assessments regarding genetic history and teratogen usage. Many managed care plans in Arizona, especially AHCCCS (Medicaid managed care) health plans are developing, or have developed, health screening and risk assessment components that focus on preventive screening services for adults and women of childbearing age. Concern is that some physicians may delay initial appointments for prenatal care until the second trimester if the woman does not complain of problems. These areas need to be more clearly identified, coordinated, and supported throughout the State, so that genetic risk screening can be a more consistent practice among all providers.

In addition, referral for genetic counseling and evaluation should be done early in pregnancy because testing of individuals or family members may be a lengthy process. Generally, if prenatal testing is available, it is done from 15 to 20 weeks in the pregnancy, but may in some cases, be available as early as the 10th gestational week in pregnancy. Therefore testing on family members must be accomplished prior to pregnancy or early in the first trimester.

ARIZONA HAS:

ADULTS WHO ARE AT RISK OF DEVELOPING PREVENTABLE GENETIC DISORDERS. Scientists working under the auspices of the Human Genome Project continue to identify genetic components to acute and chronic diseases, such as cancer, diabetes, and cardiovascular disease. The cause of these potentially life threatening illnesses is thought to be multifactorial, a combination of inherited factors and environmental agents. While the inherited factors cannot be prevented, lifestyle changes, behavioral modification, and in some cases preventive treatment may affect the manifestation or severity of the disease. Five of these diagnoses/disorders will be considered: 1) diabetes, 2) breast/ovarian cancer, 3) colon cancer, 4) hemochromatosis, and 5) adult PKU, including maternal PKU.

Diabetes

While diabetes can be inherited as a single gene disorder, either dominant or recessive, in most cases it is thought to be multifactorial (King et al., 1992). Diabetes mellitus is a heterogeneous group of disorders that have elevated blood sugar in common. Symptoms vary from unnoticed and/or mild, to ketoacidosis, retinopathy, and atherosclerosis. The high frequency of diabetes and its associated costs, morbidity, and mortality make this an important health problem in most populations (Hansen, 1995; King et al., 1992). It is the leading cause of adult blindness and amputations in the United States and a major cause of renal failure, heart attacks, and strokes (Scrivner et al., 1995)

There are two major forms of the primary illness: insulin dependent diabetes mellitus and non-insulin dependent diabetes mellitus. Diabetes can be associated with genetic syndromes, such as Prader Willi, Bardet Biedl, Cockayne, von Gierke disease, or Friedrich ataxia (King et al., 1992).

Type 1, or insulin dependent diabetes is a combination of genetic predisposition and environmental factors, thought to be viral in origin. This autoimmune process may present with the appearance of islet cell antibodies in the blood, years before observable physical symptoms of diabetes appear. To date, medical science cannot prevent this type of diabetes.

Type 2, or non-insulin dependent diabetes, appears to be primarily determined by genetic factors; autoimmunity is not involved. Multifactorial influences impact even the single gene, inherited forms. An individual who is overweight, eats a poor diet and doesn't exercise, may suffer effects of the disorder sooner and more severely than an individual with the same inherited gene who is of normal to slightly subnormal weight, eats a healthy diet and exercises regularly (Hanis, 1996).

Cancer

Everyone has a risk of developing cancer. Cancer is the abnormal proliferation and reproduction of cells within the body. Normally the body has many internal controls to limit abnormal cell proliferation, but cancer can result if these internal controls are reduced or eliminated. In genetics, the combination of genetic and environmental influences is referred to as the “multiple hit theory” (Schneider, 1995). A number of “hits” are needed to activate the cell to go out of control. One of these “hits” is an hereditary component. Other hits include environmental factors such as exposures to radiation, chemicals, occupational hazards, biologic agents (hepatitis B, HIV), and diet (alcoholic beverages, tobacco, nitrites/nitrates, animal fat, low fiber diet, etc.). The importance of genetic counseling in cancer is to review with clients the varying causes of cancer and multifactorial risks, which include inherited susceptibility and environmental factors. A discussion of an individual's risk, informed consent (including pros and cons of doing laboratory tests such as DNA), consequences of testing (such as denial of life, health or disability insurance), and possible psychological ramifications should be covered in the genetic counseling. In addition, lifestyle changes that may reduce the risk for cancer may be discussed.

Breast/Ovarian Cancer

With the identification of the BRCA1 and BRCA2 genes through the Human Genome Project comes an important step in the understanding of the biology of breast cancers. Many concerns and ethical issues, however, remain unresolved. Approximately 1 in 200 to 1 in 300 women carry the BRCA1 gene mutation (Hoskins, 1995; Biesecker et al, 1993). Not every woman with the gene will develop cancer, although her risk is 85% by the time she is age 80 (Schneider, 1995; Biesecker et al, 1993). A positive gene test does not tell when the cancer will occur, what the early stages of cancer will be, how amenable to treatment the cancer will be, whether more than one type of cancer will occur, nor which specific form of cancer will occur, if any (Schneider, 1995). Likewise, the absence of cancer does not mean the test results are wrong. Negative test results for the mutation do not guarantee that cancer will never develop.

In Arizona, breast cancer is the second leading cause of cancer deaths among women. According to the Arizona Cancer Registry, in 1995, over 3,100 women were diagnosed with breast cancer, and 570 women died from this disease.

Women who have a family history of BRCA1 or BRCA2 and are not found to have the gene can be reassured their risk is probably approaching the risk of other women in the population; generally far lower than their perceived self risk (Evans et al, 1994). Women who do have the gene would have a more specific idea of their risk. Increased surveillance in the form of starting mammography earlier and more frequently may be considered. Pelvic ultrasound may be offered to look for early development of ovarian cancer.

At the completion of childbearing, women who carry the BRCA1 or BRCA2 gene may want to investigate prophylactic mastectomies and/or removal of their ovaries. These surgical procedures

have not been shown to be totally effective in eliminating cancers in these areas. Breast and tissue near ovaries may still be present and react to the same cancer producing agents (Hulka, 1995; McPherson, 1994; Beisecker, 1993; King, 1993). There is no consensus at this time as to the "proper" method of surveillance, prophylactic measures (such as removal of breasts or ovaries), or frequency of mammography. However, genetic counseling for any woman who has a family history of such cancers may be very beneficial for her, to decide her best course of action, in consultation with her physicians.

In a study presented at the American Society of Human Genetics annual meeting in Minneapolis in October of 1995, researchers presented information on women with the BRCA1 mutation. Although knowing prophylactic surgery did not eliminate their risk for breast or ovarian cancer, women still felt that by knowing they were gene carriers they could convince their health care providers of the need for early and frequent surveillance. They felt that by having the information they were empowered and could make more informed choices in their own health care. It should be noted that cancer susceptibility testing does carry the potential for significant social, emotional, ethical, and financial issues. Testing should only be voluntary, and only after indication of increased risk, and with psychological readiness on the part of the client to accept and live with the results of the tests.

Colorectal Cancer

Colorectal cancer is the third most commonly diagnosed cancer, and the second leading cause of cancer death in the United States (Agency for Health Policy Research, 1996). In 1996, an estimated 133,500 cases were diagnosed nationwide, and nearly 55,000 people died of the disease. According to the Arizona Cancer Registry, there were 2,129 reported cases of colorectal cancer in 1995, with 830 persons dying of this disease in 1995. There are many risk factors for colorectal cancer, including age, male sex, inflammatory bowel disease, certain hereditary conditions, and a family history of colorectal cancer or adenomatous polyps. Research indicates that reductions in morbidity and mortality from this disease can be achieved through detection and treatment of the cancer in its early stages. Research indicates that colorectal cancer mortality can be reduced by 15 to 33 percent by routine fecal occult blood testing, combined with flexible sigmoidoscopy in high risk individuals (Agency for Health Policy Research, 1996). DNA testing may be done in high risk individuals.

Hemochromatosis

Hemochromatosis may be one of the most frequent genetic diseases that is inherited in an autosomal recessive mode of inheritance, with a high gene carrier frequency of 1:7 to 1:10 persons, and about 1:200 to 1:400 affected individuals (Powell et al, 1996; Washington State Dept. of Health, 1996; Rouault, 1993; King et al 1992; Lin et al., 1985). It is a disorder that is characterized by increased iron absorption and accumulation. Although hemochromatosis presumably begins at birth, clinical liver disease is seldom observed before middle age.

Diabetes, renal disease, heart disease and hepatocellular carcinoma are serious complications of hemochromatosis. These complications are completely preventable with proper treatment of the disease. Symptoms of hemochromatosis may include fatigue, weakness, weight loss, abdominal discomfort or pain, joint pain, backaches, itchiness, and frequent infections. Late signs and symptoms of the disorder may reflect the organ systems affected, such as hepatic disease, diabetes, heart irregularities, shortness of breath or chest pain. Because these symptoms are not specific to hemochromatosis, diagnosis may be very difficult. Measurement of serum ferritin, transferrin saturation test, and serum iron and/or iron-binding capacity can be done to identify individuals at risk. An abnormal result on more than two occasions is suggestive of a positive diagnosis. A liver biopsy is needed to confirm the diagnosis. Treatment involves phlebotomy, indicated by the serum iron levels (Powell et al 1996; Washington State Dept. of Health, 1996; Rouault, 1993).

The hemochromatosis gene is very closely linked to the HLA gene complex, and is associated with particular HLA genes. Therefore, the hemochromatosis gene in a family can be followed by tracking the HLA gene. Siblings of the patient with hemochromatosis who share the same 2 HLA haplotypes are also highly likely to share the 2 hemochromatosis genes, implying that they will eventually develop iron overload. A more distant relative who shares 1 HLA haplotype of an affected individual, has a 5% chance of receiving a second hemochromatosis allele (gene) from his/her other parent, because of the high gene frequency, and will then have hemochromatosis. By identifying first degree relatives with identical haplotypes to the affected individual, the risk for the disease can be increased or reduced. Identical haplotypes are highly likely to predispose the individual the hemochromatosis eventually, therefore, periodic serum iron studies should be done. Once the iron levels become high enough, then phlebotomizing is the mode of treatment. For first degree relatives who carry neither high risk haplotype, their risk will be much reduced and serum iron levels may be done less often (King et al. 1992; Lin et al. 1985).

Adult/Maternal PKU

Phenylketonuria is a metabolic disorder caused by two defective recessive genes. Phenylalanine, found in all dietary protein, is not metabolized properly. Untreated, phenylpyruvic acid builds up in the body, causing mental retardation. Formerly, dietary guidelines indicated phenylalanine restriction until the age of five. Current guidelines recommend lifetime-restricted phenylalanine diet, however, many individuals with PKU went off the special diet, at age five, and are now of childbearing age. Phenylalanine-free food products are very expensive, and are often not covered by insurance, which are deterrents to maintaining the strict diet. The diet and formula products are also unpalatable.

The maternal PKU syndrome refers to the teratogenic effects of PKU during pregnancy. These effects include mental retardation, microcephaly, congenital heart disease, and intrauterine growth retardation. Women may have classic PKU or other variants of PKU that were successfully controlled in the early life of the mother with dietary restriction. In untreated pregnancies where the mother has classic PKU with a high phenylalanine level, there is a very high risk of abnormality in the newborn, approaching 75-90% for microcephaly and mental retardation, and 15% for congenital heart disease (Levy HL & Ghavani M, 1996). The lower the phenylalanine

level, the lower the risk of the birth defect, both in pregnancies of women with PKU variants, and in classic PKU pregnancies.

Dietary restriction is critical prior to and during pregnancy to reduce the teratogenic effects on the baby. Toxic effects of phenylpyruvic acid on fetal growth and development occur when a mother with PKU is not adequately controlled by diet. These toxic effects include a high probability of mental retardation, microcephaly, congenital heart disease, intrauterine growth retardation, (Levy HL & Ghavani M, 1996).

For those women who have gone off diet, the ideal time to initiate dietary restrictions, is prior to conception or at the very latest in the first few weeks of pregnancy. If strict dietary compliance is achieved, it is possible to have normal or near normal offspring (Levy HL & Ghavani M, 1996).

Close monitoring during pregnancy is also essential, as levels fluctuate with changing metabolic needs.

B. CLINICAL SERVICES

INTRODUCTION

Regardless of the type of clinical genetic service that is provided, anyone who receives clinical genetic services should have the benefit of meeting with a genetic counselor prior to, and following the testing process. Individuals may benefit from periodic consultation with a genetic counselor as life changes occur. Genetic counseling is a comprehensive, complex, and dynamic communication process, best accomplished in a private professional setting with a Certified Genetic Counselor. In 1975, the American Society of Human Genetics and its Subcommittees on Genetic Counseling described genetic counseling (Epstein et al., 1975) as:

“a communication process which deals with the human problems associated with the occurrence, or the risk of occurrence, of a genetic disorder in a family. This process involves an attempt by one or more appropriately trained persons to help the individual or family to (1) comprehend the medical facts, including the diagnosis, probable course of the disorder, and the available management; (2) appreciate the way heredity contributes to the disorder, and the risk of recurrence in specified relatives; (3) understand the alternatives for dealing with the risk of recurrence; (4) choose the course of action which seems to them appropriate in view of their risk, their family goals, and their ethical and religious standards, and to act in accordance with that decision; and (5) to make the best possible adjustment to the disorder in an affected family member and/or to the risk of recurrence of that disorder.”

A correct diagnosis is essential to the process of genetic evaluation. The key elements in a genetic consultation/evaluation include: 1) diagnostic evaluation; 2) prognosis and management; 3) recurrence risks; 4) reproductive options (if appropriate); and 5) follow-up support. The diagnostic evaluation includes several components:

- reviewing the medical history
- reviewing the family health and pregnancy history
- reviewing the social history
- requesting previous medical reports and test results
- conducting a physical exam
- establishing a diagnosis or differential diagnosis
- requesting and completing additional tests, when appropriate
- conducting literature review, when necessary
- consulting with colleagues, when appropriate

A diagnosis is needed to be able to start the process of genetic counseling and to give parents or individuals information about the disorder. Integral to the process are the medical geneticists who are specialists in the rare disorders. Whether the disorder is a birth defect or a family history of an adult onset disease, the medical geneticists are the health professionals who are the diagnosticians. In the case of a birth defect, the geneticist may be asked to evaluate whether the etiology is primarily genetic or environmental.

The geneticists evaluate not only the system or systems affected in the child, but also a three or four generation family history, known in genetics as a “pedigree”, which diagrams the relationship between individuals regarding the health and pregnancy history. A very detailed physical exam is done, evaluating even minor anomalies. The information from the medical history, family health history (pedigree), pregnancy history, social history, medical reports, test results and physical exam is then compiled to see if a diagnosis can be made.

Genetic counselors are an integral part of the diagnostic and evaluation process. They assemble and synthesize information before the patient's initial visit, obtain and review medical records pertinent to the evaluation, conduct the family history or pedigree, and often conduct research on possible differential diagnoses. Before and after the visit with the family, the genetic counselor attempts to provide support and assistance to the family. A critical part of this support includes follow-up to see that tests recommended by the geneticist are completed, if the family chooses to obtain the additional tests. The genetic counselor also connects families to additional services and resources in the community, and is an advocate for the family when necessary.

The purpose of prenatal genetic counseling is to provide information to pregnant women and their partners. This evaluation includes an assessment of risk, such as maternal age, family history of birth defects or genetic conditions, past pregnancy history, current pregnancy information, assessment of teratogens, as well as medical history of the mother and father of the baby. A three or four generation pedigree or family tree is usually constructed. The systematic manner in which this is constructed, individual by individual, elicits more quality information about family health and pregnancy history compared to the questionnaire regarding diseases. The optimal way to obtain the family history is with both pedigree and systems review, asking questions about each system of the body as well as key words and phrases, such as “is there anyone in the family with any mental retardation, any neurologic disorders such as Alzheimer, Parkinson, or Huntington's disease?” (Congleton, 1986).

Genetic counseling prior to testing for genetic disorders should include information about health insurance issues. Possible insurance problems with positive results include cancellation of benefits, exclusions of cancer-related disorders, increase in premiums, “job lock” (inability to move to another job because insurance in the new job won't cover preexisting conditions), loss of coverage with job loss, reduction in benefits or refusal by the insurance company to entertain applications. Potential benefits of finding out one is not a carrier may be in knowing that increased cancer surveillance is not needed, thus translating to savings of health care dollars. Individuals would also know that if they did not inherit the specific mutation found in other family members, they would not pass the mutation on to their children (Schneider, 1995). Benefits may include documentation of need for increased surveillance and the earliest possible diagnosis.

**ARIZONA HAS:
A NETWORK OF GENETIC SPECIALISTS AND PRACTITIONERS**

- **PEDIATRIC GENETICISTS**
- **PERINATAL SPECIALISTS FOR PRENATAL GENETIC COUNSELING AND TESTING**

Since 1982, there have been certification exams for eligible genetics professionals through the American Board of Medical Genetics. The exams are available in Clinical Genetics, Clinical Biochemical Genetics, Clinical Cytogenetics, and Clinical Molecular Genetics. The majority of genetic counselors have at least a master's degree in genetics or nursing, and are certified by the American Board of Medical Genetics (prior to 1996), or the American Board of Genetic Counseling (from 1996 on). Physicians who are genetic specialists are usually also Board Certified in internal medicine, obstetrics/gynecology or pediatrics. Arizona's genetic service availability is focused on the treatment of infants and children, and high risk pregnant women (ADHS, 1996).

The Pediatric Genetics Program includes: the Section of Medical and Molecular Genetics in the Department of Pediatrics, University of Arizona with locations in Tucson and Phoenix. Maricopa Medical Center also has a pediatric genetics program. Genetic diagnosis and counseling are available through private providers as well as through Office for Children with Special Health Care Needs/Children's Rehabilitative Services (CRS/OCSHCN) clinics in Phoenix, Tucson, Yuma, Flagstaff, Sierra Vista, and at a number of Indian Health Service clinics throughout the state.

Four programs provide prenatal diagnosis and prenatal genetic counseling: 1) the Arizona Institute for Genetics and Fetal Medicine, in Chandler; 2) Tucson Perinatal Services, Tucson; 3) Phoenix Perinatal Associates, Phoenix, Mesa and Glendale; and 4) University of Arizona Health Sciences Center, Department of Obstetrics and Gynecology's Section of Maternal/Fetal Medicine. Although the majority of geneticists in Arizona are affiliated with the University of Arizona's College of Medicine, there are two private practice physicians that specialize in genetics in Phoenix; one prenatal and one pediatric. The University of Arizona offers a two year genetic counseling master's degree program. (ADHS, 1996).

Although adults who have cystic fibrosis and sickle cell anemia may be managed through the CRS/OCSHCN clinics, there are few genetic services in Arizona for adults. There are several facilities in Arizona that provide cancer susceptibility testing and cancer risk counseling for adults, including the University of Arizona Cancer Prevention Clinic in Tucson, the Scottsdale Mayo Clinic, and a limited number of private practice sites. The University of Arizona and Mayo Clinic programs offer multidisciplinary counseling services that involve oncologists, geneticists, mental health professionals, surgeons, dieticians, social workers, and epidemiologists.

Arizona geneticists completed a questionnaire regarding their impressions of the current status of genetic services in Arizona. The main concerns of the geneticists included the following:

- Lack of a biochemical genetics laboratory in Arizona, which can delay receipt of test results. Biochemical specimens must be sent out of state for testing, taking anywhere from 7 to 10 days. Delays in receipt of test results cause delays in treatment. Certain metabolic genetic conditions (newborn PKU, MSUD, etc.) require immediate diagnosis and early treatment to prevent and limit devastating complications. Delays in diagnosis and intervention result in poor patient outcomes, which is a critical risk management issue for practitioners.
- □ Lack of knowledge/information on the part of primary care providers (internists, family practitioners, obstetrician/gynecologists, pediatricians) about genetic disorders, appropriate referrals, familial testing, and ongoing monitoring.
- Poor payment mechanisms by managed care and private insurance companies for genetic diagnostic evaluation and genetic counseling of risks. In addition, geneticists note that there is a lack of financial incentive (and possible disincentive or penalty) for referral of patients for genetic evaluations in a managed care system.
- Lack of payment mechanism for family genetic evaluations. The individual presenting with a problem or symptoms may be covered for the evaluation; however, other family members are often not, even when an evaluation of extended family members is needed for a complete diagnosis.
- Geographic barriers due to Arizona's large size resulting in lack of physical access to local genetic testing and evaluation services.
- Limited number of diagnoses covered by CRS/OCSHCN.
- Lack of insurance coverage for low income persons (non-Medicaid eligible) and non-U.S. citizens.
- Lack of genetic services and trained providers for adult metabolic patients.
- Poor to nonexistent payment mechanisms for essential foods for metabolic disorders (such as adult and maternal PKU).

ARIZONA HAS:

A COMPREHENSIVE NETWORK OF PREVENTIVE HEALTH SCREENING SERVICES THAT PROVIDES:

- **REPRODUCTIVE HEALTH/FAMILY PLANNING SERVICES**
- **DIAGNOSTIC SERVICES**
- **PRECONCEPTION COUNSELING**

Preventive health screenings (annual physical exams, cervical and breast cancer screening, STD testing, etc.) are a covered benefit for many women in Arizona who have health insurance. AHCCCS Health Plans (Medicaid managed care plans) conduct personalized outreach to all women of childbearing age and older to encourage the utilization of these preventive health screening services. In addition, there are twenty-one reproductive health/family planning clinics throughout the state of Arizona. These clinics and health care providers offer a comprehensive array of preventive health services and diagnostic screening services, including, but not limited to: gynecological exams, cervical and breast cancer screening, sexually transmitted disease/HIV/AIDS testing, blood pressure screening, family planning counseling and education.

The reproductive health/family planning clinics provide preconception counseling to women and their partners. Clinic documentation and medical record data collection forms indicate that counselors discuss teratogen use, such as smoking, alcohol and drugs. In addition to discussing teratogen use and exposure, providers who are seeing women of childbearing age for preventive health/reproductive health/family planning services should use that opportunity to obtain additional information from the client about genetic risks. Providers can also promote the use of folic acid/folate supplementation prior to conception, to reduce the risk of having infants with neural tube defects.

A survey was developed for providers who specialize in services to treat infertility. There was a 56% response rate, (5 of 9 responding). The five respondents were interviewed, and 2 of the five respondents also completed a written questionnaire in addition to the interview. Three (60%) said that personnel (including physicians) had received inservice education or attended seminars regarding clinical genetics in the past two years. Eighty percent of the respondents (4 of 5) indicated that they assess genetic risks, but only one of the respondents had written protocols for conducting these risk assessments. Four respondents did not have M.D. or Ph.D. geneticists or genetic counselors on staff, and only one respondent noted a contractual agreement with genetic professionals. Two mentioned an informal agreement and referral process with certified geneticists.

ARIZONA HAS:

A SYSTEM OF MULTIDISCIPLINARY CARE AND SERVICES FOR CHILDREN WITH CERTAIN TYPES OF GENETIC DISORDERS.

Children's Rehabilitative Services (CRS) is a program administered by the ADHS Office for Children with Special Health Care Needs (OCSHCN). The mission of CRS/OCSHCN is to provide for, within the limits of appropriated funds, medical treatment, rehabilitation, and related support services to medically and financially qualified children/families with certain handicapped (or potentially handicapping) conditions having the potential for functional improvement through medical, surgical, or therapy modalities. CRS/OCSHCN is responsible for: monitoring the delivery of service through private contractors, keeping statistical data on the CRS/OCSHCN population, organizing and conducting field/community clinics, providing support and consultation, and ensuring overall program planning and management. CRS/OCSHCN goals are to provide quality care through early detection, prevention, and comprehensive medical treatment and rehabilitation to enrolled individuals. To establish eligibility for CRS/OCSHCN, the following three criteria must be met:

1. Specialized treatment is necessary;
2. Functional improvement is potentially achievable; and
3. Long term follow-up may be required for maximum achievable results.

CRS/OCSHCN provides comprehensive services to children from birth to age 21. Over the age of 21, only adults with cystic fibrosis or sickle cell anemia are covered. Children who are eligible for CRS/OCSHCN services who are concurrently enrolled in AHCCCS (Medicaid managed care program) have primary care services through AHCCCS and multidisciplinary specialty services coordinated through CRS/OCSHCN with the assistance of a case coordinator. Once a child has been determined to be medically eligible for CRS/OCSHCN services, there is a financial eligibility component that determines the share of cost for CRS/OCSHCN services to be borne by the family. The Appendix contains a listing of medically eligible conditions for CRS.

The CRS/OCSHCN clinics have a multidisciplinary focus, with numerous specialties being represented. Multidisciplinary teams are organized, depending on the child's needs, and the particular requirements for effective management of a disorder. For instance, the Craniofacial/Orofacial Clinic team members include: an audiologist, dentist, geneticist, genetic counselor/nurse, oral surgeon, otolaryngologist, pediatrician, plastic surgeon, psychologist, registered nurse, social worker, speech therapist, and as needed, an educator, nutritionist, and translator.

There are four CRS contractors (Regional Clinics), one in each of the communities of Yuma, Flagstaff, Phoenix, and Tucson. CRS outreach genetics clinics are held at various intervals in most counties throughout

the state. All four CRS Regional Clinics have bilingual staff and translators to serve Spanish-speaking children and their families. The CRS Metabolic Clinic treats children who are found to have a metabolic disorder as a result of newborn screening, such as phenylketonuria (PKU), maple syrup urine disease (MSUD), etc. Special formulas are necessary to manage some of the metabolic disorders. These formulas are paid for by the state through the newborn screening fees and are available through contracted pharmacies if families are enrolled in CRS/OCSHCN.

ARIZONA HAS:
AN ESTABLISHED NEWBORN SCREENING PROGRAM THAT TESTS FOR SEVEN DISORDERS

The first population based screening of newborns in the United States was begun in 1966 to detect infants with phenylketonuria (PKU) so that treatment could be instituted early to prevent mental retardation. PKU is an inborn error of metabolism characterized by a deficiency in the enzyme phenylalanine hydroxylase that results in high phenylalanine levels, which, if not diagnosed and treated early in life can result in severe, progressive mental retardation. PKU occurs in about 1 in 10,000 Caucasian births. Dietary restriction, if instituted early in life, is effective in preventing mental retardation. (Andrews et.al, 1994; Holtzman, 1991).

The purpose of genetic screening is early detection before the onset of symptoms or irreparable damage. Prerequisites for inclusion of a screening test into a newborn screening program include: (1) identifiable population at risk; (2) the disease has a high incidence and is serious; (3) the screening test is simple, safe, inexpensive, and suitable for mass processing; (4) there are options available for treatment or prevention for the disease; (5) the test reliability is high; and (6) benefits outweigh the cost (Allen DB & Farrell PM, 1996).

Today newborns are screened for several inborn errors of metabolism. Fifty states and the District of Columbia, Puerto Rico, and the U.S. Virgin Islands test for PKU and congenital hypothyroidism. Although congenital hypothyroidism is not considered an inborn error of metabolism and is hereditary in only about 10% of the cases, it fits the other criteria mentioned above.

Prior to 1979, individual hospitals in Arizona performed tests for PKU and some other conditions but there was no organized statewide newborn screening program. In 1979, the Colorado Department of Health was contracted with the state of Arizona to do the laboratory testing for the newborn screens. In 1993, the Arizona Legislature enacted legislation requiring the Arizona Department of Health Services (ADHS) to develop and administer a formal Newborn Screening Program. The Newborn Screening Program conducts newborn screening tests for seven (7) disorders, as well as confirmatory testing for hemoglobinopathies. These particular

disorders have been selected because their detection and treatment in the newborn period is effective in preventing severe morbidity or mortality.

The disorders screened are:

1. Congenital hypothyroidism
2. Phenylketonuria (PKU)
3. Galactosemia
4. Homocystinuria
5. Maple syrup urine disease (MSUD)
6. Biotinidase deficiency
7. Hemoglobinopathies

Revenue from the newborn screening program test fees provides funding for specialized formulas that are necessary with some of the disorders. Infants that require special formulas because of metabolic disorders covered throughout the newborn screening program are covered through the CRS/OCSHCN Program.

The following table illustrates the incidence of newborn screening disorders identified from 1994 through the most recent data available in 1997:

Disorder	1994	1995	1996	1997
Congenital Hypothyroidism	13	38	27	32
MSUD	1			
Biotinidase Deficiency	1		2	0
Phenylketonuria (PKU)			4	3
Hyperphenylalanemia			2	5
Galactosemia		1		
Homocystinuria				1
Sickle Cell Anemia	10	9	7	4
Annual Births Reported	70,896	72,368	76,741	75,094

**ARIZONA HAS:
A NETWORK OF DETECTION, TREATMENT, AND MANAGEMENT FOR CHILDREN AND
ADULTS IDENTIFIED WITH SICKLE CELL ANEMIA.**

In 1972 Arizona Sickle Cell Anemia Program (ASCAP) was established as a result of a mandate by Arizona Public Health Control laws ARS § 36-797.41 and ARS § 36-797.42 "...to make every effort to detect sickle cell anemia, a heritable disorder...". Addendums to the 1972 statutes were added to appropriate monies for pediatric (1980) and adult (1983) sickle cell clinics with comprehensive medical rehabilitation treatment programs in Phoenix and Tucson.

In 1986, hemoglobinopathy testing was added to the Newborn Screening profile for all Arizona infants. Testing for hemoglobinopathies in Arizona is done by a method known as isoelectric focusing. Testing is also available without charge to any individual who believes he/she is a member of a susceptible population. ADHS offers hemoglobin electrophoresis testing through a contract with Good Samaritan Medical Center in Phoenix. State funds and Maternal and Child Health Block grant monies are used to pay for sickle cell counseling and screening services.

The emphasis of the Sickle Cell Anemia Program in Arizona centers on the identification, diagnosis, and treatment of newborns with sickle cell disease, to prevent physical impairments and deaths due to sepsis and bacterial infections. Once an infant is identified with an hemoglobinopathy through newborn screening and confirmation testing, family members may receive additional testing, and are given training in how to prevent active disease in their children. Through this program there is also a critical opportunity to identify carriers of the disease and provide counseling, supportive therapies and treatment.

In 1995, 10 infants were identified with sickle cell disease and 1498 infants with hemoglobin traits. In addition, ASCAP identified two adults with sickle cell hemoglobinopathy and followed up in 85 children and adults identified as having positive hemoglobin traits. Counseling was provided to 787 families with positive hemoglobin screens.

ASCAP also offers clinics for children and adults. The clinics are administratively funded through the Office of Children with Special Health Care Needs (CRS/OCSHCN). Referrals for enrollment and treatment are coordinated by ASCAP. Sickle cell counselors are trained by the State Sickle Cell Program Manager to educate individuals and families about any abnormal hemoglobins. Counseling is provided by professionals and lay community workers who contract with the Sickle Cell Disease Association of America, Arizona Chapter, which was incorporated in 1977. This community-based organization provides advocacy and support services to individuals and families diagnosed with sickle cell disease. There is one full time staff person whose position is funded through a contract with ASCAP and corporate donations.

Individuals with sickle cell anemia may be seen in the CRS/OCSHCN clinics as children and may continue those services as adults. Sickle cell disease and cystic fibrosis are the only disorders covered by CRS/OCSHCN for the adult population.

ARIZONA HAS:

ONLY A FEW PROVIDERS WITH GUIDELINES ADDRESSING CRITERIA FOR PROVISION OF, OR REFERRAL TO, GENETIC SERVICES

Questionnaires were sent to eight specialty clinics in Arizona: 1) The Cystic Fibrosis Clinic, Phoenix Children's Hospital; 2) Hemophilia Clinic, St. Joseph's Hospital, Phoenix; 3) Muscular Dystrophy Clinic, Phoenix; 4) Neurofibromatosis Clinic, Phoenix Children's Hospital; 5) U of A Muscular Dystrophy Clinic, University Medical Center, Tucson; 6) Hemophilia Clinic, University Medical Center, Tucson; 7) Myelodysplasia Clinic, Los Angeles Children's Hospital; and 8) Shriner's Clinic, Phoenix. Several of these questionnaires were conducted in person or by phone. All eight clinics responded to the questionnaire. The largest clinic serving the most clients (782 visits in 1994) was the University of Arizona Medical Center Muscular Dystrophy Clinic. Clinic sessions range from one half to one day in length. Specialty clinics hold between 6 and 107 clinic sessions per year. Seven of the eight specialty clinics surveyed (88%) use a multidisciplinary approach to service delivery. A total of 25 different types of health professionals provide services in these clinics. Geneticists were available in 2 of 8 (25%) clinics. In general, few providers who were surveyed during the needs assessment indicated that they had guidelines or criteria available to guide their decision-making in the assessment and/or referral of persons suspected to have a genetic disorder.

Only one clinic had written guidelines that address genetic counseling. Contractual agreements with a geneticist (M.D. or Ph.D.) occurred in 3 of 8 (38%) of these clinics. Arrangements for consultation with geneticists are variable. Some clinics refer patients to geneticists, some do not refer patients for genetic consultation, and some provide their own genetic counseling services. Seven of eight respondents (88%) offered DNA carrier testing.

ARIZONA HAS:

INCONSISTENT ACCESS TO GENETIC SERVICES FOR CHILDREN

Preventive health care services for children are available throughout Arizona and accessible through primary care physicians, clinics, and managed care delivery systems. Although the access to preventive health and screening services for children is widespread, the access to genetic specialty services is not consistent, especially among children who are not eligible for low income or federally funded programs.

Medicaid (AHCCCS) Health Plans in Arizona must participate in the federal EPSDT Program (Early and Periodic Screening, Diagnosis, and Treatment). Children from birth through age 20 enrolled in AHCCCS are to receive age appropriate medical screening exams according to an established periodicity schedule. EPSDT services provide comprehensive health care, (as defined in A.A.C. R9-22-213, and 42 CFR, 441.58) through primary prevention, early intervention, diagnosis and medically necessary treatment of physical and behavioral health problems for eligible AHCCCS members under 21 years of age. EPSDT also provides for all medically necessary services to treat or ameliorate physical and behavioral health disorders, a defect, or a condition identified in an EPSDT screening. Components of the EPSDT screening include:

1. A comprehensive, developmental, nutritional, medical and social history.
2. A comprehensive, unclothed physical examination.
3. Appropriate immunizations according to age and health history.
4. Laboratory tests (including blood lead level assessment appropriate to age and risk).
5. Health education.
6. Appropriate dental screening.
7. Appropriate vision and hearing testing.

Pediatric genetic services in the state are provided by board certified medical geneticists and genetic counselors at the University of Arizona in Tucson and in Phoenix, as well as by a board certified medical geneticist/genetic counselor at Maricopa Medical Center. The University of Arizona genetics staff in Tucson and Phoenix contract with ADHS to provide outreach services in a variety of centers throughout the state for CRS/OCSHCN.

Challenges to Genetic Service Access for Children and Families

There are many challenges to obtaining genetic services for Arizona's children. These challenges relate to intended or unintended barriers such as:

- lack of appropriate referrals to genetic services by primary care providers
- medical eligibility
- financial eligibility
- insurance coverage and benefit package
- problems in coordination of care
- lack of parent and provider information about available services and resources
- lack of transportation

Genetic screening and testing services for children must usually be accessed through a primary care physician, especially in a managed care system. There is concern that these referrals to genetic

services may not be encouraged by the primary care providers. The needs assessment did not determine the number of EPSDT or primary care provider visits that result in genetic service referrals.

Referral to genetic services is variable, depending on the health system involved. According to one geneticist, Indian Health Service (IHS) appears to refer to pediatric geneticists most consistently of all groups in Arizona. IHS has a contract with the University of Arizona for the provision of genetic health care services in seventeen clinics across the state. In addition, the focus of IHS is particularly attuned to genetics as a public health issue. Thus, IHS may be more likely to refer patients for appropriate evaluation and counseling. Other groups and managed care organizations may be more focused on individual health issues and primary health care. By seeking to control costs by limiting referrals for subspecialty care, according to one geneticist, the result may be “inadequate and inaccurate diagnosis of children with multiple handicaps and therefore, inadequate and insufficient treatment being provided.” According to another geneticist, primary care physicians may order unnecessary or inappropriate tests, many of which are expensive, causing a delay in proper diagnosis and treatment.

Geneticists surveyed agreed that IHS genetics clinics should continue. The most common reason for referrals from IHS varies from site to site, but overall the most common reason for referral is known or potential teratogenic exposure during pregnancy. Approximately 85% of referrals to the IHS clinics are to rule out fetal alcohol syndrome. Geneticists did not have information about the statewide genetic services utilization patterns, or provider's referral patterns to genetic services.

Some of the challenges to obtaining genetic services through CRS/OCSHCN relate to the established medical eligibility criteria for the services. There are children with handicapping genetic conditions that are not covered by the CRS/OCSHCN program. The most common genetic disorders not eligible for CRS/OCSHCN services are: Down syndrome (uncomplicated, such as without congenital heart defect); fetal alcohol syndrome (FAS) and other disorders due to teratogenic exposures; other chromosomal disorders; Marfan syndrome; Turner syndrome (uncomplicated, in other words, no cardiac or renal anomalies); fragile X syndrome; Prader-Willi syndrome; and skeletal dysplasias (without surgical or orthopedic bracing needs).

Other barriers to receiving genetic services through the CRS Program may include families who are not financially eligible. In order to be enrolled in CRS/OCSHCN, a child must first meet the medical eligibility criteria. Once the medical eligibility for the child has been established, the financial eligibility for services is established. Families who qualify for AHCCCS (Medicaid) are financially eligible to receive CRS/OCSHCN services with no co-payment. For families who are not eligible for AHCCCS, a sliding fee scale is established that considers the family income and resources. The Children's Hospital Program, funded through Arizona Tobacco Tax dollars, provides primary care and specialty services for children. Families who meet the medical eligibility criteria, but not the financial eligibility criteria through either AHCCCS or CRS/OCSHCN sliding fee scales, are still eligible to receive services through the CRS/OCSHCN network (to the extent that services are available from the physician network). Families are required to pay the full price for the services (100% co-payment) and then bill their own insurance separately, if available.

Coordination of care for children and families who need genetic services is sometimes difficult. Because of the demand for genetic services in general, the CRS/OCSHCN clinic sites and private offices may have a lengthy waiting period between a referral and the appointment for genetic services. Because the clinics are sometimes held infrequently, (i.e., bi-annually, quarterly, etc.), the timing of the clinic may not always coincide with the child's needs, thus requiring some families to travel long distances for more timely services. Families who move before their scheduled clinic appointment date may not get connected to services in their new location. In many cases, this is due to parent's lack of information about the continued need for the services, or lack of information about resources and networks in an unfamiliar area.

Some families may have difficulty with obtaining transportation to appointments. Public transportation is not available in many areas of the state. If a referral has been approved for genetic evaluation services through AHCCCS, medically necessary transportation (e.g., bus tickets, taxi service, etc.) is a covered benefit, and is provided upon request through the health plan, when the member/family does not have other options for transportation.

Most of the geneticists surveyed believed that not all children in Arizona who needed genetic services were receiving adequate services. Some of the barriers geneticists mentioned included distance from the clinic site, transportation problems, lack of payment by insurance, lack of follow through by managed care for referrals. One geneticist commented that some children are not being referred to genetic services due to the primary care physician's lack of knowledge of the condition and/or available services.

Birth defects occur in all racial groups, although some defects are more common among certain groups. Based on current data available, there is an over representation of Native Americans in Arizona's program, even when fetal alcohol syndrome is excluded, according to one geneticist who responded to a needs assessment survey. "...This is because private genetic referrals (either through insurance companies, HMOs, or AHCCCS) are grossly under-represented. Thus it is my feeling that individuals with private health insurance receive much worse genetic health care than do children covered by CRS/OCSHCN or the Indian Health Service."

Overall, geneticists reported that most CRS/OCSHCN families are returning to the clinics at the appropriate intervals for follow-up visits. Families who are reminded of the clinic appointment by letter, postcard, or telephone have a higher "show rate". In addition, where patients have identified transportation problems, follow-up visits are more likely to occur in areas where social services can assist with arranging transportation. Where public health nurses are involved in the IHS clinics there is a higher "show rate." Where insurance doesn't cover the visit, or if HMOs, AHCCCS, or private insurance are involved, the children may not return for follow-up visits. The cost of the genetics visit, if borne by the family, appears to be a deterrent to returning for the follow-up visit.

ARIZONA HAS:

LIMITED INFORMATION ABOUT THE AVAILABILITY OR UTILIZATION OF ADULT GENETIC SERVICES

Arizona's genetic service network centers around identification and management of genetic disease in children. Genetic conditions often become apparent early in life, or are associated with developmental problems that are recognized at an early age. The component of the Arizona adult population that is commonly identified for use of genetic services are at-risk pregnant women, or couples who are experiencing multiple miscarriages or infertility. Very little is currently known about the numbers of adults in Arizona that are referred for genetic counseling and evaluation because they have a family history of a single gene disorder. According to the geneticists surveyed, referrals of adults for genetic services are infrequent; thus, it is likely that Arizona's adult population is significantly underserved.

The need for genetic services for adults will continue to increase. Several environmental and social factors may account for this need:

1. Increasing age of the population, and longer life span.
2. Many genetic disorders do not fully manifest themselves until adulthood, or with age.
3. New technological advancements in the identification of adult onset disease genes, and their impact in the development of disease.
4. Improved medical management of pediatric genetic disorders, helping children survive into adulthood.
5. Persons with PKU must maintain lifetime dietary restrictions, especially women of childbearing age.

An additional needs assessment needs to be developed, implemented and analyzed. Because the majority of adult onset diseases has a genetic component, this becomes a daunting task.

C. LABORATORY SERVICES

ARIZONA HAS: AVAILABLE TECHNOLOGY FOR PERFORMING GENETIC SCREENING TESTS

Maternal Serum Screening

Maternal serum screening is a noninvasive laboratory test for pregnant women to obtain information about fetal development. Initially developed to identify risk for neural tube defects (NTD), maternal serum screening in routine obstetrical care provides critical information for pregnancy management and risk assessment, and the potential for early prevention or treatment. Population screening identifies from a given population a smaller group of patients who are at risk of a disorder. When identified, this smaller population is offered specific diagnostic tests, and subsequent treatment if necessary. Screening should be voluntary, and the pregnant woman should be counseled about test risks and limitations prior to taking the blood test.

The alpha fetoprotein (AFP) test began as the “single marker” test, and detects approximately 80% of infants with neural tube defects and 30% of infants with Down Syndrome. The addition of other serum “markers” to the AFP test, such as estriol, and beta HCG (human chorionic gonadotropin), has been found to be useful in identifying patients at risk for other fetal structural malformations and chromosomal anomalies. This “multiple marker screen” (or triple screen) will still detect 80% of neural tube defects, but will increase the detection rate of Down Syndrome to 60% and will also be positive 30-40% of the time in detecting trisomy 18. The AFP test may also be elevated with underestimated gestational age, multiple pregnancy (more than one baby producing AFP), fetal death, abdominal wall defects (such as gastroschisis or omphalocele), renal defects, fetal teratoma, and can indicate an increased risk for abnormal pregnancy outcome even with a normal fetus.

Multiple marker screening tests are done from 15 to 17 weeks after the first day of the last menstrual period. Results are usually available within 5 to 10 days. Women then have the option of diagnostic studies (i.e., ultrasound and/or amniocentesis), accompanied by genetic counseling. The multiple marker screen has an approximate 10% false positive rate. A “false positive” result means that the screening test indicates a potential problem, but subsequent diagnostic tests (via ultrasound, chromosomes and AFP testing on the amniotic fluid) are normal. Recent studies indicate an increased risk for preterm birth, fetal death or other complications in pregnancies with abnormal multiple marker screening on the mother's blood, but normal ultrasound and amniotic fluid AFP and chromosomes. There are also false negative results, in which the multiple marker test is normal, but spina bifida is in fact, present. It is important to note that in approximately 20% of spina bifida cases, the spinal defect is covered with skin, thus no spinal fluid leaks into the amniotic fluid, and the MSAFP may be normal. An ultrasound may detect this defect if the infant is positioned optimally.

An abnormal result achieved during the diagnostic testing phase gives the parents options for management, including but not limited to: preparation of the family for a baby with problems, possible change in obstetrical management, and preparation for delivery of a baby with NTD by Caesarean section. Most cases of NTD (90-95%) occur to couples with no apparent family history.

Couples who have a previous child with an NTD will usually be offered high resolution ultrasound and amniocentesis based on the recurrence risk of 2-3%. That risk may be different if there is more than one family member with an NTD, or is associated with a single gene or chromosomal disorder.

The maternal serum testing, whether it is the single marker AFP or the multiple marker test, is not a diagnostic test; it is a screening test. A normal result does not ensure the baby will not have a birth defect, nor does a positive test indicate a baby will have a birth defect. A positive test does indicate a patient has a level of risk high enough to warrant additional diagnostic testing, such as ultrasound and/or amniocentesis.

The College of American Pathologists (CAP) provides critical laboratory proficiency testing for MSAFP tests. The American College of Medical Genetics strongly supports regulations for quality control of any laboratory involved in AFP testing. Typically, AFP test results are reported as multiples of the median (MOM) which is a statistical convention by which different laboratories can compare their results, regardless of the variations in the lab or the gestational weeks in the pregnancy.

Questionnaires about laboratory processes were sent to nine laboratories, with four responses (44%). Three of the four labs responding sent their maternal serum screening specimens to another lab for analysis. The one lab which performed the analysis of maternal serum screening specimens received both in-state and out of state samples. The number of samples received by labs ranged from 210 to 12,000 in 1994 and 144 to 13,000 in 1995. Two of the labs responding to the questionnaire offered single (AFP), double (AFP & HCG), and triple marker (AFP, HCG & estriol) screening. One lab offered single and triple marker screening. The fourth lab offered only triple marker screening. Costs in 1996 ranged from \$30 to \$60.50 for the AFP only, \$48 to \$91 for the double marker, and \$78 to \$179.90 for the triple marker.

The one laboratory that did AFPs in house was accredited by the College of American Pathologists, Arizona Department of Health Services, New York State Department of Health Services, Florida and Pennsylvania State Clinical Lab Licensure. Staff included an ABMG certified Ph.D., genetic counselor, and consultant board certified physician geneticists available.

In general, the labs to which maternal screening tests were directed depended on the contract specified by the payor. AHCCCS health plans reimburse for maternal serum screening. One plan reimbursed for AFP only, two reimbursed for AFP and AFP + HCG (respondents were both unsure

if triple screen was reimbursed) and ten plans reimbursed for all three methods of maternal serum screening (AFP only, AFP + HCG, and AFP + HCG + estriol).

Of the 14 AHCCCS plans 13 routinely reimburse for maternal serum screening. Of those 13, all 13 reimbursed for AFP only; 12 of 13 for AFP + HCG and 10 for the triple marker screen (AFP + HCG + estriol).

Reimbursement is dependent on what the provider orders, and/or the particular reimbursement arrangement of the obstetrical provider. Seven of 13 (54%) stated the MSS screening is part of the obstetrical package. Three of 13 (23%) stated these services are not part of the obstetrical package, and two of 13 (15%) said it depends on the provider.

Cytogenetics

A questionnaire was sent to eight laboratories with four responding (50%). The four labs received a total of 3,529 specimens for calendar year 1995 with the following breakdown of samples:

CVS	196	(6%)
Amniotic fluid	1,143	(32%)
Lymphocytes	1,066	(30%)
Bone marrow/tumor	839	(24%)
Products of conception	285	(8%)

Two of the labs reported that they send specimens to reference labs (one lab sending specimens to three different reference labs). The other two labs evaluate specimens at their own facility. Both labs who perform the testing are accredited by CAP (College of American Pathologists), participate in proficiency testing, and maintain clinical and billing databases. One responded they do not obtain or maintain any pregnancy outcome information on tested cases. The other did not respond. One did FISH (fluorescent in situ hybridization).

ARIZONA HAS:

NO LABORATORY THAT PROVIDES DNA TESTING OR QUANTITATIVE ANALYSIS FOR THE CONFIRMATION OF METABOLIC DISORDERS

Arizona currently sends quantitative amino acid studies out of state for confirmation of newborn screening disorders. Because of the unavailability of this testing in Arizona, test results can be significantly delayed, causing delays in final diagnosis and treatment. There are currently no laboratories in Arizona that are doing quantitative amino acids or molecular testing, according to the medical geneticists surveyed. Molecular testing is very specific, and not every molecular lab can perform every test. In most cases, providers who need to request a particular test must contact a number of sources to find a laboratory that will perform the specific test. Helix is an online national database of laboratory tests that are available to be performed at specific laboratories. On line or paper medical literature searches also identify locations for testing.

D. HEALTH PROMOTION AND PREVENTIVE HEALTH SERVICES

The ultimate goal for the science of genetics is the prevention of disease. Primary prevention of congenital malformations is a relatively new concept for public health.(Khoury, 1996) Historically, public health prevention efforts involved education about teratogenic medications and rubella immunizations. Optimally, women should not take medications during pregnancy. Exceptions can include women with epilepsy who are controlled on anticonvulsant therapy. Discontinuing medication could have devastating effects to both mother and fetus. Alternatives for medical management should be promoted, such as monotherapy (one drug) or reduced dosages of medications. Changes in drug therapy should optimally be instituted prior to conception. The risk for adverse effects of an uncontrolled serious medical condition (i.e., seizures, resulting in lack of oxygen to the fetus) may be deemed to outweigh the risk of possible teratogenic effects in such circumstances.

Unfortunately in Arizona, many women experience unintended pregnancies, so the opportunity for pre-pregnancy modification of therapies or behavior may be moot. (Brown & Eisenberg, 1995.) Unintended pregnancies are defined as pregnancies that are mistimed (but wanted at some future time) or unwanted. According to the Arizona Women's Health Survey conducted in 1993, approximately 36 percent of women 18-44 years old who were married or living in consensual union had experienced an unintended pregnancy (the last pregnancy within the past five years). Approximately 62% of women age 18-44 who were previously married or those previously living in consensual union living together had experienced an unintended pregnancy (Arizona Women's Health Survey, 1993). Preconception planning can help women to time the quantity and spacing of their children, and can improve maternal and infant outcomes.

ARIZONA HAS:

A PUBLIC HEALTH SYSTEM THAT SUPPORTS PRIMARY, SECONDARY, AND TERTIARY PREVENTION PROGRAMS, AND INCORPORATES FAMILY SUPPORT, ADVOCACY, AND HEALTH PROMOTION CONCEPTS

“Prevention” activities can be categorized in three levels. Primary prevention includes activities, treatments or practices undertaken to prevent the development or occurrence of an illness. An example of primary prevention is the prevention of fetal alcohol syndrome by avoiding alcohol during a pregnancy. Another example of primary prevention includes reduction in the occurrence of neural tube defects by taking folic acid prior to conception and throughout the first few months of pregnancy. Folic acid has been clearly demonstrated to reduce the incidence of structural developmental anomalies in early fetal development, especially neural tube defects. Secondary prevention refers to preventing the complications of a disorder through appropriate, timely intervention or treatment. Examples of secondary prevention include prevention of mental retardation in hypothyroidism with thyroid replacement therapy, and treatment of phenylketonuria (PKU) or maple syrup urine disease (MSUD) with special formula and dietary restrictions. Tertiary prevention occurs when serious complications of a disorder can not be alleviated, but there may be benefits to certain treatments. For example, children with Down syndrome have varying degrees of mental retardation. These children may benefit from early intervention programs to help them achieve their highest potential function. Also, because children with Down syndrome are at an increased risk, it is recommended they be screened for hypothyroidism. (Khoury, 1996)

ARIZONA HAS:
LIMITED AVAILABILITY OF, AND ACCESSIBILITY TO, PRECONCEPTION GENETIC SERVICES

In Arizona, many women and their partners do not obtain preconception genetic screening services. We believe the health care community considers genetic services only valuable after birth, or after the woman has already entered prenatal care. Furthermore, few insurers cover preconception genetic screening or testing services.

ARIZONA HAS:
EMERGING PROGRAMS FOR CONSUMER AWARENESS REGARDING FOLIC ACID SUPPLEMENTATION AND AVOIDANCE OF TERATOGEN USE
Neural Tube Defects/Folic Acid

One emerging area of research is the link between folic acid supplementation and the reduction in the occurrence of neural tube defects. Neural tube defects (NTD) are a group of birth defects involving the cranium and spinal cord, such as spina bifida, anencephaly and encephalocele. They result from the failure of proper closure of the spinal column which occurs from 16 to 28 days after conception. Although there are some circumstances that may increase the risk of having a child with an NTD (such as some seizure medications used during pregnancy), the cause of most NTDs is unknown. There is speculation that folate metabolism, controlled by as yet undiscovered genes, influences the action of folate on the developing fetus.

There have been studies linking maternal use of folic acid/folate with a reduced incidence of NTD (Khoury, 1996; CDC, 1992; Milunsky, 1989). It is recommended that all women of childbearing age, whether pregnant or not, consume 0.4 mg of folic acid daily. Higher dosages are recommended for women who plan to become pregnant, who have had a child with an NTD, or who have a family history of NTD in either parent. The folic acid supplementation must be started very early in the pregnancy or optimally, preconceptually, because the fetal spinal cord develops before most women know they are pregnant. If a woman waits until she confirms her pregnancy at six weeks or later, that is too late, and the development of the spinal cord is complete.

In 1995, the Arizona Department of Health Services (ADHS) formed a committee to promote folic acid/folate supplementation. The Committee is comprised of ADHS personnel from the Offices of Women's and Children's Health, Nutrition, Epidemiology and Chronic Disease, Children's Rehabilitative Services, Children with Special Health Care Needs, genetic counselors, consumers, and other health care providers. The Folate Committee collaborates with the Arizona Perinatal Trust and the March of Dimes, who have also encouraged the use of folate. The Folate Committee developed a program plan to promote folic acid use, particularly in women of childbearing age. The Committee designed an educational brochure for statewide distribution and an educational consultant has been hired.

Teratogens

Another area where birth defects may be prevented is through reduction or avoidance of exposure to teratogens. Teratogens are environmental agents, chemicals, or drugs that can cause birth defects. When consumed during pregnancy, alcohol is considered a teratogen, resulting in fetal alcohol syndrome (FAS) or fetal alcohol effects (FAE). These birth defects are completely preventable by abstinence from alcohol. Alcohol use during pregnancy is believed to be highly understated; providers may elect not to question a woman about her alcohol consumption, and women, if asked, may not answer accurately.

The infertility clinics surveyed all provided information to clients on smoking, drinking and drugs and folate/folic acid supplementation. Three of these clinics also provided additional information on other teratogenic substances. All clinics surveyed thought that there should be a statewide teratogen information and/or counseling hotline.

ARIZONA HAS:

PUBLISHED INFORMATION ABOUT GENETIC SERVICES AND PROGRAMS IN ARIZONA

Newborn Screening Education

From 1970 through October, 1994, Arizona contracted with Colorado to perform the laboratory testing for all newborn screening specimens. No organized statewide program was in operation during this time frame. Hospitals could choose which newborn screening tests were done and to which laboratory the samples were sent. Initially, only PKU was tested, then other tests were added to the newborn screening panel done by Colorado.

In 1993 the Arizona Legislature enacted a statute requiring every newborn to be screened. A comprehensive program was developed with MCH Title V funds, and administered by the Arizona Department of Health Services (ADHS) Office of Women's and Children's Health (OWCH). A contract for laboratory services was offered for bid and was won by the ADHS State Laboratory. The law requires for this contract to be competitively bid every four years. Follow up is carried out by ADHS/OWCH Newborn Screening Follow Up Program. Initially, in 1994 and 1995, a series of regionalized educational sessions were offered in Flagstaff, Phoenix and Yuma hospitals. Additional training has been offered on request or in response to identified problems. The Newborn Screening Program will hire an educational consultant to provide technical assistance throughout the state. The Newborn Screening Program has also completed the *Newborn Screening Program Guidelines* which has been distributed to all Arizona hospitals, physician offices, and laboratories that handle newborn screening specimens.

The Mountain States Regional Genetic Services Network (MSRGSN) has developed the revised *Newborn Screening Practitioner's Manual*, available upon request. It identifies the disorders that are screened on the Newborn Screening panel in each of six states in the MSRGSN (Arizona, Colorado, Montana, New Mexico, Utah and Wyoming). The manual also includes information on the disorders screened, and state laws governing newborn screening.

Hemoglobinopathy Education

The Arizona Department of Health Services (ADHS) provides education through brochures, educational videos, films, and didactic presentations. ADHS also contracts with the Sickle Cell Disease Association of America, Arizona Chapter, to provide statewide public education.

The Council of Regional Networks for Genetic Services (CORN) has developed *Guidelines for Follow-up of Carriers of Hemoglobin Variants Detected by Newborn Screening*. The basic points are:

- (1) Ideally, education about newborn sickle cell testing should be provided to families during prenatal care well in advance of the time of delivery;

- (2) A mechanism should be in place in State Newborn Screening Programs so that all results of sickle cell newborn screening can be made available to the parents of all infants who are tested;
- (3) Parents of all infants who are detected to be carriers of hemoglobin variants should be offered appropriate education, counseling, and testing;
- (4) Individuals who counsel should have appropriate training and credentialing in order to insure the highest quality of services for families of carriers detected by newborn screening;
- (5) Newborn screening programs should have a mechanism for monitoring and assessing the approaches to, responses to, and costs of providing carrier education and counseling services. The Sickle Cell Disease Association of America is currently working on two projects regarding national certification of hemoglobinopathy counselors, and development of policy and guidelines on trait follow up and counseling. It is recognized that there is a need to provide professional credentials, transferable from state to state.

Genetic Services Education and Awareness

General education about genetic services is accomplished in a variety of ways. A booklet “Genetic Services in Arizona”, published by ADHS, is in its third edition and is widely distributed throughout the state. This document provides information about the types of services that are available, brief descriptions of the services, and sources and locations for receiving the services. In addition, there is a listing of other sources of information for people affected by genetic disease or birth defects. This publication is geared to health care practitioners, rather than parents or consumers. This publication was distributed to consumers upon request, and was well-received.

Genetics professionals often refer patients to Arizona's Pilot Parents and the Emily Anderson Learning Center for information, advocacy, and support. Geneticists and genetic counselors speak about genetics issues in community forums. Most of the geneticists believed the time they had available was more often spent educating health professionals than the lay public.

A two-year project conducted by the ADHS Office for Children with Special Health Care Needs analyzed laws, rules, and policies for six major programs in the state that impact children with special health care needs. The team was comprised of representatives from ADHS Office for Children with Special Health Care Needs, Children's Rehabilitative Services, ADHS Behavioral Health, DES Division of Developmental Disabilities, AHCCCS, Arizona Long Term Care Program (ALTCS), and Arizona Early Intervention Program (AzEIP), Children's Action Alliance, and consumer/parent groups. Findings of the Legislative/Policy Analysis team indicated that eligibility rules and laws for these programs were not necessarily in conflict, but that the implementation policies of the various organizations caused fragmentation, confusion, and limitations to service access for children with special health care needs. Parent consumers recommended creation of an easy-to-read user friendly reference for parents with children suspected of needing specialized, multidisciplinary, interagency services, diagnoses, or treatments. CRS and MSRGSN have developed a brochure to include questions that parents and families may wish to ask of their genetics providers.

Results of the parent/consumer forum held on March 3, 1995, indicated that parents wanted additional accurate information about genetic services to be readily available to the public through media, doctors offices, and other community networks. Parents wanted access to specific information regarding genetic disorders, including treatment and coping approaches, via new technology (World Wide Web, medlines, etc.). Parents also wanted more training for primary care health professionals in all disciplines about genetic services, conditions, and referral options.

ARIZONA HAS:

AVAILABILITY OF GENETIC EDUCATION PROGRAMS IN ARIZONA COLLEGES OF

MEDICINEOne of the geneticists surveyed noted that recent graduates from area residency programs are more aware of the need for genetic evaluations and the availability of such services. This geneticist stated, "older physicians in the community are less aware of genetics in general, as well as the importance of genetic diagnosis and counseling for their patients. This subgroup of physicians is in need of further education and information about the importance and availability of genetic services." Overall, geneticists in Arizona strongly recommended improving the breadth and scope of genetic services awareness and education that is provided to the general public and to health care professionals and medical practitioners.

Arizona geneticists do routinely lecture about genetics to pediatric, family practice and OB/GYN residents in the University of Arizona College of Medicine and at community hospital-based residency programs. Geneticists regularly present at Grand Rounds, speak at conferences, and provide periodic educational programs to health care workers in outlying areas of the state, including Indian Health Service staff. All geneticists interviewed acknowledged that they were providing education to health professionals, but that they had not yet focused on public education and awareness activities.

At this time, Arizona needs more complete information about the specific genetics education provided in nursing schools and paraprofessional training institutions. Arizona also needs better information about educational offerings to health professionals, including continuing medical education.

ARIZONA HAS:
**A PROGRAM FOR IMPROVING ACCESS TO, AND UTILIZATION OF,
GENETIC SERVICES FOR ARIZONA'S HISPANIC POPULATION**

The Arizona Department of Health Services and the University of Arizona Health Sciences Center, Genetics Department, received a SPRANS grant (Special Projects of Regional and National Significance) from the Maternal and Child Health Bureau of the U.S. Department of Health and Human Services. This grant is entitled "Improving Access to and Utilization of Genetic Services to Arizona's Hispanic Population". This project expands genetics clinics in the state, and teaches community lay health workers in ADHS's Health Start

Program basic human genetics. To encourage prevention, early identification and treatment, the grant supports paraprofessional curriculum development and implementation, Spanish language training for health professionals, and culturally competent service delivery to this growing segment of Arizonans.

ARIZONA HAS:
A NATIONALLY RECOGNIZED PROGRAM FOR REVIEW OF CHILD FATALITIES, WITH THE MISSION OF REDUCING PREVENTABLE DEATHSADHS has a Child Fatality Review Program whose mission is to reduce preventable child (including infant) fatalities through systematic, multi disciplinary, multi agency, multi modality review of child fatalities in Arizona; through interdisciplinary training and community based prevention education; and through data driven recommendations for legislation and public policy. In essence, the Child Fatality Review Program is a data-driven model for implementing strategies to prevent childhood deaths. Multidisciplinary teams meet to assess circumstances surrounding the child fatality in their community, review the data related to these deaths and recommend public policy and private action that can prevent child deaths in the future. Included in these deaths are preventable deaths caused by genetic disorders.

Theoretically, this program may, over time, collect data about children who have died as a result of a preventable genetic disorder. This program has the potential to provide information to support the development of additional tests for the newborn screening program, and other birth defects prevention programs for Arizona. The Arizona Birth Defects Registry is another databased surveillance program and is described on page IV-49.

E. SOCIAL, LEGAL, AND ETHICAL ISSUES

“Ethics is a method of analysis enabling us, temporary intruders on human agonies, to play our critical role in such a way as to make our intervention appropriate and our behavior consistent” (Frederick R. Abrams). Bioethical issues are not unique to genetics, or to medicine. Who decides what the patient's treatment entails? Should the physician as expert, tell the patient what the course of treatment will be, or does he/she give alternatives, pros and cons of each, and facilitate, rather than dictate? The principles most regularly applied in biomedical ethics are autonomy, non-maleficence, beneficence, justice, veracity and fidelity. Autonomy involves the right of the patient to choose (and for some, the right of the physician to choose). It encompasses a respect for the patient, the family and their values. It requires open-mindedness, and a critical reflection on one's own and others' approaches to care of patients, self scrutiny of potential or real inadequacies and/or biases (Am Board of Pediatrics--Medical Ethics Subcommittee). Non-maleficence is to do no harm, and beneficence, meanwhile, places a responsibility on us to do good; non-maleficence and beneficence may at times conflict. Although a fundamental principle in biomedical ethics, justice is one of the most troublesome. Different approaches to the question, “is this action just?” will elicit opposing responses, i.e., justice for whom, the individual? Society? Justice refers to what is right, treating all cases alike, to be fair. Veracity is telling the truth, and fidelity is the keeping of contracts or covenants.

What does the geneticist/genetic counselor do who has counseled a couple and finds the results of the tests reveal non-paternity? How far does the health care provider go when finding out a pregnant woman drinks copious amounts of alcohol, or uses cocaine? Does the health care provider have the right to hospitalize and thus isolate the woman who has PKU who does not adhere to her diet, knowing the baby will, with a high degree of certainty, be mentally retarded and have other birth defects?

When an individual receives information about their genetic makeup, the information may be relevant to other family members and future generations in the family, as well. Who tells the other members of the family they are at risk? Is it up to the health care provider or the patient?

Genetic testing often identifies “risks” related to an individual or their offspring that do not pose a threat to the individual's health, but merely identify potential risks for their reproductive outcomes. Genetic testing may also be used for diagnostic purposes, which may precede any symptoms. This testing may require decisions regarding treatment and management based on an expected or predicted prognosis. Also, genetic testing may reveal a condition for which there is no medical cure or treatment. Genetic conditions, if recorded in a patient's record, could result in an insurer refusing to cover that condition, or charging an unacceptably high premium for coverage.

ARIZONA HAS:

NO PLAN FOR ADDRESSING SOCIAL, LEGAL, AND ETHICAL ISSUES FOR GENETICS

Respect for persons should be the basis of all genetics services. Not only is this an ethical approach, it is the most effective strategy for communication and care. Providers who regard consumers as partners in their own care demonstrate respect for the other's intelligence and personhood. Such providers should listen to consumers and take their questions and comments seriously. The Consumer should be regarded as an equal active partner in his/her treatment.

Genetic conditions are likely to have a profound impact on the whole family, including both genetic relatives and relationship by adoption. The integrity of the whole family must be considered, even if only one member comes for genetic counseling.

Some Basic Principles Underlying Genetics Services:

1. Respect for people with genetic conditions by helping them to live and to make reproductive and lifestyle choices according to their own values.
2. Equal and fair allocation of public resources to those who need them most.
3. Freedom of choice, with the woman as final decision maker in reproductive matters, including abortion decisions.
4. Voluntary decision making, with avoidance of coercion by government, society, health care providers, or other such groups.
5. Respect for human diversity, and respect for people whose views are in the minority.
6. Respect for the consumers' basic intelligence and their views of their situations, regardless of level of education or knowledge about genetics.
7. Increased education about genetics for public, health professionals, teachers, and clergy.
8. Close cooperation with organizations for persons with genetic conditions.
9. Prevention of unfair discrimination or favoritism based on genetics in employment, insurance, schooling, or other areas.
10. Teamwork with other professionals through a network of referrals.
11. Inclusion of consumers as respected members of the team to the maximum degree possible.
12. Use of nondiscriminatory language that respects individuals as persons. New England Regional Genetics Group 1996

Much needs to be done in this area to educate consumers, and health care professionals. Community based forums should be held for an exchange of information and ideas, about priority issues in this area. It is not uncommon that health care providers decide what the consumers need without asking them. In a study by Lindsay Middleton, nurse geneticist at the National Institute of Health, consumers were asked to identify the five most important and five least important psychosocial issues in the area of prenatal and pediatric genetics. The findings were surprising to many geneticists:

Five Most Important Issues--Prenatal Clients

1. To discuss how I will learn about the results
2. To learn about the accuracy of the testing
3. To learn about the risks of the prenatal testing
4. To find the chance of the genetic condition occurring in me, my child and other family members
5. To find out what to expect when having prenatal testing

Five Least Important Issues--Prenatal Clients

1. To talk about my or my partner's pregnancies
2. To learn about pregnancy termination options
3. To discuss my feelings about pregnancy termination
4. To decide if I should have another child
5. To discuss reproductive options

Five Most Important Issues-- Pediatric Clients

1. Medical treatment/management available
2. To find out what is wrong
3. To learn coping skills
4. To find out if the condition can be cured
5. To find out the chance of the condition occurring in me or my child

Five Least Important Issues--Pediatric Clients

1. My relationship with partner/spouse
2. To plan future pregnancies
3. To discuss availability of prenatal testing
4. To talk about my or my partner/spouse's pregnancies
5. To discuss alternative reproductive options

Five Least Important Child Impact Issues--Pediatric Clinics

1. Child's relationship with siblings
2. Child's ability to have children
3. Getting or keeping health insurance
4. Getting or keeping life insurance
5. Child's relationship with friends/neighbors

**ARIZONA HAS:
NO GUIDELINES ADDRESSING GENETIC TESTING IN CHILDREN**

The following guidelines for genetic testing of children take into account the increasing respect for minors' autonomy in the overall context of medical care (Wertz et al., 1994; American Society of Human Genetics, 1995). Testing falls into four general categories depending on its benefits for the child or adolescent:

1. *Testing for conditions for which treatment or preventive measures are available.*

Examples are familial polyposis coli, where removal of the colon in the early teenage years is necessary to prevent cancer, and familial hypercholesterolemia, where diet and medication reduce cholesterol levels. For disorders where proven methods of treatment or prevention exist, testing of minors should proceed according to consent guidelines established for other necessary medical treatments (Nicholson, 1986; Holder, 1989). Testing should be offered at the earliest age when health benefits accrue, and not before that time.

2. *The test has no health benefits for the minor, but may be useful to the minor in making reproductive decisions now or in the near future.*

This is one of the more common reasons for requests for testing. Examples are carrier testing for autosomal or X-linked recessive disorders (e.g., cystic fibrosis or Fragile X), or pre-symptomatic testing for adult onset disorders (e.g., Huntington disease). It may be useful to a minor of reproductive age to know his/her genetic status. In such cases, the minor should be the primary decision maker. Professionals should probe to discern whether the minor is acting on her/his own behalf or is merely carrying out parental wishes without actually desiring to be tested.

Testing done to inform reproductive decision making should be limited to situations where the potential risk to a minor's offspring is high (e.g., for dominant disorders). Ordinarily, carrier testing for autosomal recessive disorders will not be warranted unless either the minor or the minor's partner has a family history of the disorder. There may be exceptions for some well defined, high risk ethnic groups, such as Ashkenazi Jews who are at risk for Tay Sachs disease.

3. *There are no medical benefits and no current reproductive benefits from testing, but parents or the minor request it.*

Examples include parental requests for cystic fibrosis carrier testing of their children or Huntington disease testing on children who are well below reproductive age or who are not contemplating reproductive activity in the immediate future. Sometimes children or adolescents themselves request testing. They may do so in order to consolidate a developing self identity regarding work or parenthood. A test showing that the minor does not carry the gene would undoubtedly relieve anxiety at this age, especially if a member of the immediate family had symptoms of the disorder. The benefits and harms of a test showing the presence of a gene, however, are more difficult to weigh.

There is no research evidence about optimal timing or the benefit of testing. For some, it may be less traumatic to find out about genetic risks to offspring early on, before self image is developed. For others, it may be better to wait, and others might benefit most from not knowing until adulthood.

Testing in the absence of medical benefit or current reproductive decision making is the most ethically problematic category of testing. There are no real parallels; testing is not “necessary” medical care and does not relate to reproductive rights.

The age at which the emotional maturity required for consent appears is highly variable and also depends on the seriousness of the genetic disorder. Often it will be advisable to defer testing until adulthood. If no clear benefit(s) exist, parents should restrain their desire to know.

The professional should act as the minor's advocate by insuring that both minor and parents are aware of potential harms and by thorough testing of the minor for cognitive, emotional, and moral maturity, using referrals if necessary. Ordinarily competence should not be assumed. Testing in cases without medical or reproductive benefits requires careful evaluation.

Sometimes a request, whether from a parent or a minor, must be rejected as serving no useful purpose and possibly leading to harm. Such decisions that override parental autonomy may be necessary in order to prevent harm and to preserve a minor's future autonomy, which should be the paramount consideration. Actions that place parental autonomy above all other concerns may lead to harm (Thomasma 1983; Brett & McCullough 1986; Engelhardt 1982). For example, a parental request to test a three year old for adult polycystic kidney disease or a seven year old for predisposition to familial Alzheimer disease provides no medical benefit to the child and may lead to stigmatization.

4. *Testing is carried out solely for the benefit of another family member.*

This occurs frequently in DNA linkage analysis, where several members of a family, linking both individuals with the condition and without the condition, must be tested in order to find out whether a particular individual (or a fetus) has a gene. Sometimes small children must be tested in order to enable their parents to use prenatal diagnosis in the next pregnancy. Such testing has a clear medical benefit, but not to the individual tested.

Thorough counseling of both minor and family (including siblings) should precede testing, in order to assess the inner strength of all concerned. In determining a minor's capacity to request or consent to testing, professionals could use accepted criteria: reasonable outcome of the minor's decision, rational reasons leading up to the decision, and presence of a fairly stable set of values that will continue into the future. Standardized psychological tests for anxiety and depression are inadequate to predict responses to knowledge relating to one's health 20 to 40 years later.

Professionals should find out precisely how each child or adolescent views the cause of a genetic disorder, and should take the child's conceptions seriously. They should remember that well informed, articulate children or adolescents, or adults for that matter, are not necessarily coping well with the prospect of illness, or perceived illness. Professionals need to examine their own motivation in providing information, and should proceed carefully in correcting distorted ideas and in dealing with egocentric or magical thinking (NERGG, 1996).

Minors should have the "negative right" of not knowing about their genetic status if they so desire (Beauchamp and Childress, 1994). Autonomy includes both the right to know and the right not to know one's genetic status.

Adoption

Adoptive parents may be more interested in risks faced in childhood than in risks for adult onset disorders. They should know the results of the newborn screening and of any other newborn tests done (on the basis of the child's family history) for disorders that may appear in the future.

The same approaches suggested for parents' biological children should also apply to adopted children and children awaiting placement for adoption (Morris et al., 1988). Testing a child for un-treatable adult onset disorders prior to adoption makes the child into a commodity undergoing quality control. If testing is carried out for the benefit of a biological family member, the consent of the adoptive parents should be required, and anonymity of all parties should be preserved.

Disclosure of Test Results to Children

In situations when genetic testing could show whether there may be a possible genetic condition but there is no helpful clinical intervention, parents have three options: 1) do not disclose to the child the existence of the disorder in the family; 2) disclose the disorder's existence but allow the child to decide about testing when he/she reaches the appropriate stage of development; and 3) disclose and also try to have the child tested. Option 2 provides the greatest respect for the child or adolescent as a person and is the most ethically acceptable.

Testing of Adults with Genetic Conditions

The problem with many adult onset genetic disorders is that measures undertaken in the name of prevention may have no proven efficacy. Tests should fall into the "ordinary" or "necessary medical care" category only if treatment or prevention has an acknowledged and proven efficacy. In all other

cases, the treatment associated with a test should be presented to the family as experimental, and the consent/assent procedure requirements for testing should follow a research protocol.

Ethical and Social Issues with the Human Genome Project

The National Center for Human Genome Research was established by Congress in 1988. The specific aim was to map the estimated 50,000 to 100,000 genes in the human genome and determine the sequence of the 3 billion base pairs of nucleotides that make up human deoxyribonucleic acid (DNA), the basic genetic material (Antonarakis, 1991; Watson 1991). A portion of the funding has been set aside to develop a program to anticipate, analyze, and address the ethical, legal, and social implications (ELSI) of the new advances in human genetics that the human genome research will make possible.

The goals of the ELSI component are to improve professional and public understanding of these implications through research and education, to stimulate informed public discussion of the issues, and to develop policy options intended to ensure that genetic information is used for the benefit of individuals and society. The ELSI research program has focused on several high priority areas raised by the most immediate potential applications or consequences of genome research:

- ☐ ethical issues surrounding the conduct of genetics research;
- ☐ responsible clinical integration of new genetic technologies;
- ☐ privacy and fair use of genetic information; and
- ☐ professional and public education about these issues.

The opportunity of acquiring genetic information with new techniques found by investigators through the Human Genome Project has implications for other social practices beyond the scope of health care policies. With new techniques which can use small samples of tissues, such as blood, then amplify the information (with a test called polymerase chain reaction, PCR), public health programs such as newborn screening and samples collected for personal identification, such as the military or the criminal justice system, can become rich repositories of genetic information about individuals and their families. The privacy of such information, who "owns" the information, who can use the information and for what purpose, are questions that remain unanswered.

Currently there are no uniform standards governing the privacy of these de facto DNA databases (Andrews, 1990). The ELSI Program has facilitated groups consisting of legal, philosophical, and scientific studies to develop the knowledge base necessary for policy making in this area. Private and public sector genetic databases and repositories have requested assistance in improving their privacy protection. Legal challenges are not resolved, and are not even totally delineated.

Forensic scientists have been using DNA typing for personal identification of convicted felons and the building of a national database. As DNA typing has entered the courtroom, questions have been raised about reliability, both in terms of methodological standards and the population statistics that underlie it (Billings, 1992).

Another important social policy need identified was improved protection against employment discrimination on the basis of genetic information. Both the NIH and the American Medical Association (AMA) have condemned the exclusionary use of genetic testing by employers (AMA, 1992; Healy, 1992). Bills prohibiting exclusionary genetic testing by employers have been introduced in California, Iowa, and Arizona.

Curbing health costs has become a primary goal of employers, as that is the most significant area of increasing costs of employee benefits. Genetic risk information has been used by life, health and disability commercial insurance as part of their underwriting process. Laws have been enacted to attempt to prevent discrimination, but most do not prevent the increasing of premiums. A new law was recently enacted in Arizona that prevents genetic discrimination with respect to life and disability insurance (see Appendix H).

Legal Issues in Genetics

Many legal questions raised in genetics can be handled by precedents set by other types of medical practice. However, the science and practice of medical genetics extends far beyond other areas of medicine (Andrews, 1987). Ethicist/attorney Alexander Capron stated “a technology that might peer into our genes and even transform them cuts to a deeper level of psychological significance than do others in bio-medicine” (Capron, 1985). Identifying a genetic defect or disease in an individual is much different from identifying an infectious disease. The genetic constitution is an integral part of an individual, and a genetic problem viewed as a defect in one's own makeup. Individuals, who unknowingly are carriers of a genetic disorder that arises in their child, feel guilty, though they had no way of knowing before the child was born. Risk for carrier status extends to relatives, and is unique to medical genetics.

The doctor-patient relationship has traditionally been considered confidential and privileged. A physician did not have an obligation to reveal medical information to anyone but the patient. As health insurance companies came into prominence, they insisted on the right to the test results if they had paid for the testing. The insurance companies applied actuarial analysis to rate individuals with disorders, who theoretically, over time, could be more costly than individuals rated as “healthy”, or at normal risk. In truth, the latter group was merely a cohort for whom they lacked information about potential genetic negative outcomes. Companies have often refused to take on the health insurance costs for certain genetic conditions, or rated the policies so high individuals could not afford health insurance.

In addition, a physician, paid by a company to do pre-employment physicals or pre-insurance physicals, did not have to disclose any adverse findings to the “patient”. Times have changed and now, the physician who is working for the individual's employer or insurance company has a duty to disclose to the applicant, employee, or insured, any assessment that the individual is susceptible to, or suffering from, a genetic or other disorder (Coffee vs. McDonnell Douglas Corp., 8 Cal. App. 3d551, 105 Cal, Rptr. 358. 502 P. 2d 1366 [1972]; Andrews, 1985).

Whether medical geneticists are obligated to recontact patients when new information arises has not been clearly delineated in the courts. When an individual is found to carry a significant genetic disorder, who is responsible for notifying his relatives?

Where are the boundaries of confidentiality? What responsibility do relatives owe to patients? In linkage studies, the DNA on a child or fetus may not be understood unless it is compared with parents and other relatives. Can a relative be forced to donate blood to participate in such a study? Currently, relatives do not legally have to participate in such studies (NERGG, 1996; Andrews, 1985).

Family members have a moral obligation to warn (or ask the provider to warn) relatives who may be at risk. In rare cases when an individual refuses to disclose or to permit disclosure of information to relatives at genetic risk, the genetics professional faces a conflict between maintaining individual confidentiality and preventing harm to the relatives. This issue is highly controversial, and it is important to distinguish between ethical obligations and legal obligations. In regard to ethical obligations, the U.S. President's Commission (an interdisciplinary body of scientists, ethicists, legal experts, and other professionals) developed the following guidelines in 1983. The genetics professional may be permitted to override individual confidentiality if the following four conditions are met:

- a. All efforts to persuade the individual to disclose information voluntarily have failed.
- b. There is a high probability of harm to the relatives (including future children) if the information is not disclosed, and there is evidence that [if disclosed] the information would be used to prevent harm.
- c. The harm averted would be serious.
- d. Only genetic information directly relevant to the relatives' own medical status would be revealed. Information relevant to the individual should remain confidential.

The President's Commission believed that overriding confidentiality should be legally permissible, but did not argue that there was a legal obligation to override confidentiality in these cases. As of 1995, no court cases have been brought against providers who overrode confidentiality while following the President's Commission guidelines. In 1993, the Committee on Assessing Genetic Risks of the Institute of Medicine (IOM) reaffirmed the President's Commission guidelines.

"The committee recommends that confidentiality be breached and relatives informed about genetic risks only when attempts to elicit voluntary disclosure fail, there is a high probability of irreversible or fatal harm to the relative, the disclosure of the information will prevent harm, the disclosure is limited to the information necessary for the diagnosis or treatment of the relative, and there is no other reasonable way to avert the harm." (Institute of Medicine, 1993, p. 278.)

F. DATA COLLECTION AND ANALYSIS

ARIZONA HAS:

NO STATEWIDE, COORDINATED PLAN OR EFFORT TO:

- **COLLECT GENETIC SERVICE DATA**
- **MEASURE AND MONITOR CLIENT OUTCOMES**

Arizona's health programs are dependent on the use of data and information for planning and decision-making. Although there is an abundance of data being collected by the health care industry, there is little coordination or interface between each of the systems. Because referrals to genetic services may be initiated from several different sources, it is also difficult to identify or track these services. Furthermore, health care providers do not have an organized, coordinated method for addressing genetic service utilization or outcomes monitoring. Several existing data collection and monitoring systems are described in the paragraphs that follow.

Arizona Birth Defects Monitoring Program

The Arizona Birth Defects Monitoring Program (ABDMP) is a statewide, population-based surveillance program that began in 1986. Information had been gathered from 1979 through 1983, but a more formal process began and decisions were made as to how and what data was going to be collected. Trained ABDMP staff collect data from 71 reporting sources: 64 hospitals; two centers providing genetic services; four CRS clinics; and the state Office of Vital Records.

Sources of data at hospitals include the disease index, labor and delivery log, nursery log, newborn intensive care log, pediatric log, and pathology/autopsy log. Not all sources are available at each hospital. An abstract of the medical record is completed for each reportable case.

In addition to the hospital sources, Certificates of Birth, Death (up to one year of age), and Fetal Death that indicate a birth defect are reviewed by ABDMP personnel and matched against cases listed in the birth defects registry. Medical records are requested from the reporting hospital on those children not previously identified from other sources and, if the condition(s) reported meet the case definition, pertinent information is abstracted for the registry. If the nature of a defect diagnosed in the first year of life is more precisely diagnosed later in the child's life, and this information is contained in the chart at the time of the review by the ABDMP (which can occur 1 to 3 years after the fact), then the precise diagnosis is used.

The abstracts of cases identified from multiple sources are compared, merged, and added to the birth defects registry. Inconsistencies, differences and/or conflicting data are resolved before being entered into the ABDMP system.

ABDMP staff assign a six digit classification code to each defect. The classification system used in Arizona is a CDC modification of the British Pediatric Association (BPA) Classification of Disease, a system similar to the International Classification of Disease (ICD), and collection is for ICD-9-CM Codes 740.00-759. The system is hierarchical: the more digits, the more precise the diagnosis. ABDMP staff always code the birth defects registry data at the most precise level possible.

As of August, 1997, birth defects for the years 1986 through 1991 have been abstracted, and the 1990 report has been issued. The process of abstracting records for all disorders proved to be very time consuming, with limited resources (abstractors), limited funds, and a subsequent backlog in data abstraction of several years. In 1996, ADHS decided to discontinue data collection regarding the more difficult diagnoses (mainly affecting the cardiac murmurs), in order to improve the timeliness of overall data abstraction. This change did not impact collection of information regarding 47 diagnoses recommended by the Center for Disease Control and Prevention (CDC). More diagnoses were and continue to be abstracted, but only the 47 CDC-recommended diagnoses are identified in the annual report.

Council of Regional Networks for Genetic Services (CORN)

The Council of Regional Networks for Genetic Services (CORN) is a federally funded program to improve the quantity, quality, and availability of cost effective genetic services nationwide. The overall purpose of CORN is:

1. To promote communication through information sharing
2. To develop a uniform system for data collection and analysis
3. To develop a system for quality assurance in specialized laboratory and clinical services
4. To initiate educational efforts for health care professionals and consumers
5. To advocate for methods of reimbursement for genetics services
6. To advocate for policies relating to the provision of genetics services

The major accomplishments of CORN have been:

1. The establishment of a national and regional system of data collection.
2. The development and distribution of several national guidelines position papers and statements.
3. The information sharing and networking that have been promoted through the regional networks and CORN between the public health sector and genetic services providers (Meaney 1992).

CORN is divided into ten regions which include the 50 states, Puerto Rico and Virgin Islands. Arizona is part of the Mountain States Regional Genetics Network.

Mountain States Regional Genetics Services Network

In response to a request from the Genetic Services Branch of Health and Human Services to identify the use of genetic services, the Mountain States Regional Genetic Services Network (MSRGSN) Data Committee developed a six state regional communal database.

The MSRGSN data set is unique because data are collected by specific patient and visit for each reporting site in the region. Data are collected on two sets of patients; prenatal and general genetics (children, adult males and non-pregnant women). Reporting sites are university based genetics centers, private practice sites and public health sites. The database has evolved over the years in a number of ways. There are two data forms (prenatal and general genetics). The forms gather considerable information on sociodemographics, procedures, diagnostic tests and test results. In 1996, the MSRGSN Data Committee approved a move to create a Windows-based data entry and reporting system. This allows reporting sites easier access and more flexibility entering and reporting their own data.

Evaluation of the data system is conducted twice a year by the Data Committee. The data forms are being changed to collect data that is relevant and accurate. The diagnostic codes are being changed to the British Physician Codes and technical support is available to the reporting sites through a contract with a private computer company.

Office for Children With Special Health Care Needs (OCSHCN)/Children's Rehabilitative Services (CRS)

CRS/OCSHCN maintains a systematic and continuous collection of data regarding CRS members. CRS/OCSHCN providers collect data and information about the services and treatments rendered to CRS/OCSHCN members, using standardized coding and definitions. Each member "encounter" is recorded and entered into the database. Query reports are then generated to monitor the utilization and quality of CRS/OCSHCN services. Indicators have been developed to monitor the quality of services and CRS/OCSHCN contractor performance. Evaluation and analysis of this information is used to improve the program by: identifying needs, trends, and patterns; educating providers and agencies about CRS/OCSHCN services; identifying potentially undeserved areas; and developing policy to guide program operations.

Arizona Cancer Registry

The Arizona Cancer Registry (ACR) was established by legislative mandate in 1980 to determine incidence patterns in the State of Arizona. The ACR is a centralized data system designed to collect, manage, and analyze information on cancer patients.

Through 1991, the ACR was a hospital-based system whereby patients with cancer were identified upon a visit to a hospital, whether as an inpatient or an outpatient. Information about the case was subsequently reported to ACR on a voluntary basis by the hospital's tumor registry, or was collected by ACR registrars at the hospital.

In June of 1988, Arizona Revised Statute §36-133, was amended by adding a new section that mandated the reporting of cancer cases. Rules to ensure reporting of all cases went into effect as of January 1, 1992.

The rules require reporting from hospitals, clinics, and doctors. The ACR has increased its efforts to become a population-based surveillance system and is striving for complete and accurate ascertainment of incidence cases in Arizona. The ACR's first population-based (complete reporting) year will be the 1995 diagnosis year. When that year is closed out in May of 1997, at least 95% of cases for that year will be reported. The surveillance system will address issues such as: identification of high risk populations, geographic distribution of cancer, identification of high risk areas, the identification and development of intervention and prevention programs.

Arizona Health Status and Vital Statistics

The Arizona Department of Health Services publishes an annual update of information on the health status of Arizona residents. The report includes statistics on pregnancies, births, abortions, stillbirths, reportable diseases, deaths, marriages, divorces, and populations of the state. One of the fundamental functions of ADHS is to identify health issues specific to the State and to develop, implement and evaluate programs and interventions that control and prevent adverse health effects. The Vital Statistics report assists in identifying critical information on topics ranging from prenatal care and pregnancy outcomes to ethnic differences in morbidity and mortality.

The first chapter deals with reproductive and perinatal health, characteristics of women who became pregnant, factors related to the course of their pregnancies, and the status of pregnancy outcomes. The second chapter is focused on trends and patterns in mortality. It compares the annual age adjusted profile of leading causes of death between Arizona and United States residents. The third chapter deals with morbidity, levels of disease in the population. Chapter four focuses on Arizona's standing in 1995 in regard to *Arizona 2000* and selected major national health objectives for the year 2000. The health indicators are organized around five subject areas: maternal and infant health, reduction of sexually transmitted and vaccine preventable diseases, prevention of injuries, age-related objectives, and mortality from chronic diseases.

Newborn Screening Program

Arizona's Newborn Screening Program utilizes the Neometrics Case Management System. This database is designed to handle the various activities and responsibilities associated with tracking and managing newborn screening data. Laboratory sample data is loaded daily into the system. The system automatically initiates a comprehensive array of follow up tasks, based on laboratory results. For example, the system generates initial and second follow up letters, prepares patient reports, provides patient history information, and coordinates other follow up activities.

Child Fatality Review Program

The Child Fatality Review Program has a comprehensive, detailed, confidential data base for collecting and summarizing data regarding deaths reviewed through the program. Data is analyzed to provide information for use in planning prevention programs to reduce the incidence of childhood deaths.

AHCCCS

The Arizona Health Care Cost Containment System (AHCCCS) maintains a Prepaid Medical Management Information System (PMMIS). AHCCCS contracts with health plans for the provision of health care services for Medicaid eligible Arizonans. Each of the health plans is required to report encounter data to AHCCCS. An encounter is a record of patient services.

Infertility & Perinatology Services

Two of the five infertility providers responded that they have clinical databases. Two of the three perinatology services have a medical information system that tracks procedures and outcomes.

V. Recommendations for Arizona Genetic Health Services

V. GENETICS HEALTH SERVICES PLANNING GUIDE

This chapter outlines suggested standards, options and recommendations for providing and improving genetic health services in Arizona, including recommendations specific to the needs of preconception and prenatal clients, children and adults. Throughout this planning guide, we have assumed that efforts to improve genetic services for preconception and prenatal clients are also considered to be primary prevention efforts directed toward children. Several key needs were identified:

1. The need for health care providers to be aware, informed, and educated regarding genetic services. Health care providers include health care and social service providers at all levels, physicians, nurses, hospitals, insurance companies, and adoption agencies. Providers need to realize that they collectively have tremendous potential for impacting the genetic health of Arizonans. Early intervention, age appropriate risk assessment, appropriate referral to genetic testing and diagnostic services, and early treatment, all make a difference.
2. The need for community awareness and information about birth defects, especially those that are preventable, and the availability of genetic services. Community awareness activities should be culturally and linguistically appropriate, include the general public, and should also be targeted to all levels of schools and community organizations.
3. The need for individualized patient information and education regarding teratogen use, the benefits of preconception health planning, healthy behaviors during pregnancy, and availability and accessibility of genetic services.
4. The need to develop and support collaboration, cooperation, and service continuity among health care service networks, governmental agencies, and community agencies related to genetic services and the prevention of birth defects.
5. The need to improve data collection, integration and reporting capabilities related to: genetic risk assessment, genetic services, and incidence of genetic conditions.
6. The need to enhance Arizona's laboratory/service network and availability for conducting certain types of genetic tests.
7. The need to refine components of the needs assessment (conduct a Level II needs assessment) to assess the need for additional services, such as Teratogen Information and Education Service, and adult genetic services.
8. The need for Arizona to address "medical necessity" and insurance issues related to the availability and accessibility of genetic services. Preconception and genetic counseling, genetic evaluation, and multi-disciplinary adult clinics are usually not covered, because they are "not medically necessary".
9. The need to inform the Arizona Legislature of the requirements of the genetics community.

PARTICIPANTS AND PROGRAM INVOLVEMENT

Any plan developed to address genetic service issues should be a collaborative process; one that includes all system stakeholders, and is the result of sustained, integrated partnerships. The following agencies and programs (not an all-inclusive list) may be involved in assisting in the development and implementation of specific plans for genetic services in Arizona:

ADHS Behavioral Health Program
ADHS Child Fatality Review Program
ADHS Community Health Nursing Program
ADHS Folate Coordinator
ADHS Health Start Program
ADHS Newborn Screening Program
ADHS Newborn Intensive Care Program
ADHS Office of Nutrition
ADHS Office for Children With Special Health Care Needs
ADHS Office of Planning, Evaluation, and Statistics
ADHS Reproductive Health/Family Planning Program
ADHS Sickle Cell Program
ADHS Substance Abuse Program
Adoptive Parents
AHCCCS
AHCCCS Health Plans
Arizona Early Intervention Program
Arizona Cancer Registry
Arizona Department of Education
Arizona Department of Insurance
Birth Defects Registry
Children's Rehabilitative Services (CRS/OCSHCN)
Chronic Disease Prevention
Commercial/private insurers
Community and advocacy agencies
Community Health Nursing
Consumer support groups
Department of Economic Security (DES)
DES Office of Evaluation
Genetic counselors
Genetic support groups
Geneticists
Health care provider community

Health service educational institutions
Health care professionals
Healthy Mothers/Healthy Babies Program
Healthy Families (DES)
Hospitals
Laboratories
Legislators
Managed care plans
March of Dimes
Mountain States Regional Genetic Services Network
Newborn Intensive Care Program
Obstetrician and Gynecology Groups
Office for Children With Special Health Care Needs (OCSHCN)
Physicians
Physician Organizations
Schools (public and private)
State, local or county government
State Laboratory
The Arizona Perinatal Trust
University of Arizona Genetics Program
University of Arizona College of Medicine
WIC (Women, Infants and Children)

GENETIC HEALTH SERVICES PLANNING GUIDE ORGANIZATION

Recommendations for genetic health services in Arizona are derived from sixteen “gold standards”. The gold standards were developed as high level “ideals” for a statewide genetics program. Some of the standards may be more difficult to achieve than others, but incremental progress for any gold standard would greatly improve the quality of the program. The standards were based on the prioritization of the key needs assessment findings.

These general recommendations are intended to be used to guide genetic service planning efforts in Arizona. The document is intended to be used as a stepping stone for further discussion, dialogue, and strategic planning for genetic services. We hope that there will be development of additional, specific projects and priorities, and ongoing evaluation of plans and processes.

ARIZONA NEEDS:

1. FEWER CHILDREN BORN WITH PREVENTABLE BIRTH DEFECTS

- **ALL WOMEN OF CHILDBEARING AGE TO BE AWARE AND INFORMED ABOUT PREVENTABLE BIRTH DEFECTS, TERATOGENS, AND MATERNAL ILLNESSES THAT CAN AFFECT PREGNANCY**
- **ALL WOMEN OF CHILDBEARING AGE TO TAKE A DAILY FOLIC ACID SUPPLEMENT**
- **MORE PEOPLE AVOIDING UNHEALTHY BEHAVIORS AND TERATOGEN EXPOSURE**

Babies in Arizona are born with many different types of birth defects. For a number of these disorders, such as neural tube defects and fetal alcohol syndrome, there are ways to prevent or reduce the occurrence of these disorders in the newborn population. Arizona can continue to focus resources on preventing certain types of birth defects through early intervention, education, and anticipatory guidance. It is also possible to mobilize resources toward minimizing untoward effects and sequelae of many “non-preventable” genetic disorders. Primary prevention of birth defects in children can be accomplished through avoidance of behaviors or teratogens that put individuals at risk for producing offspring with genetic defects. In conjunction with primary prevention efforts, ADHS can also address secondary prevention through the development of systems and support for early identification, treatment, and prevention of complications of non-preventable birth defects. Tertiary prevention is aimed at minimizing the negative impact and effects of a genetic disorder to the child and family (e.g., early intervention programs for children with developmental delays). (Khoury, 1996)

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Develop a community-based provider training and education program.**

We suggest additional training for health care providers in detection of preconception and prenatal risks (such as teratogen exposure and maternal illnesses), and methods of intervention to reduce preventable birth defects. Provider education in the prevention and/or control of maternal illnesses can be a critical factor in reducing adverse pregnancy outcomes. These provider education programs can focus on prevention of neural tube defects, and other birth defects caused by teratogen exposure (to alcohol, cocaine) or maternal illnesses (maternal diabetes, rubella, maternal PKU).

Arizona's genetics provider community can also provide information and support through continuing education programs related to early identification and prevention of birth defects caused by teratogen exposure. Training and support activities can also be encouraged relative to identification of at-risk pregnancies, and in identification of non-pregnant women who are also at-risk.

▪ **Promote prevention of fetal alcohol syndrome (FAS) and alcohol related birth defects.**

The estimated incidence of fetal alcohol syndrome (FAS) in the population is approximately one in one thousand births. During 1986 through 1990, the years for which data are available through the ABDMP, there were 89 cases of FAS reported (ADHS; 1990 Birth Defects Monitoring Program Report, Epidemiologic Report Series 1997:4) In these years there were 327,669 live births and fetal deaths in Arizona, for a rate of 0.27 FAS cases per 1,000. In 1995, when there were 72,883 live births and fetal deaths (ADHS Vital Statistics, 1995), approximately 20 cases of FAS would have been expected using the 1986-1990 rate. Since the ABDMP ascertains cases up to 1 year of age only, this number probably falls short of the actual number of cases born in 1995. The latter is likely to be understated, as many women may not be accurately reporting their drinking habits.

In a survey of geneticists, one reported that nearly 85% of Indian Health Service (IHS) referrals to genetic services were to rule out (FAS) fetal alcohol syndrome.

Despite the efforts of public health programs and the health care community, many women of childbearing age are continuing to need treatment for alcohol and substance problems. Arizona needs more information in this area to be able to better target its prevention strategies and action plans where they will have the most benefit.

▪ **Promote folic acid supplements for all women of childbearing age.**

Arizona policymakers and planners should consider periodic assessment of needs to identify appropriate strategies for improving folic acid utilization. These strategies and actions may include, but are not limited to:

- Summarizing available research on preconception folic acid supplements.
- Developing and implementing a Children's Information Center (CIC) survey on folate and vitamin supplements.
- Reviewing Center for Disease Control and Prevention (CDC) written guidelines for folic acid supplementation.
- Facilitating the development of clinical practice protocols for folic acid supplementation.
- Taking an inventory of currently available information regarding folate intake.
- Continuing to monitor folate usage through use of a survey.

- ***Inform and educate the public about preventable birth defects.***

Arizona has developed aggressive anti-smoking campaigns aimed at teenagers, in an effort to promote community awareness about a known carcinogen. Arizona should consider similar campaigns aimed at women of childbearing age about the dangers of alcohol use, especially during pregnancy. Efforts can be closely coordinated with behavioral health and substance abuse centers. In addition, an educational campaign is currently being developed around folic acid supplementation, and its role in reducing the occurrence of neural tube defects. Specific recommendations include:

- Expanding and enhancing Teratogen Information and Education Service (TIES). We recommend exploration of options for full funding of a TIES service, including a full time genetic counseling position, physician support, and funding for developmental materials and resources.
- Working with the provider community, county health departments and consumers to identify and promote community-based, culturally appropriate information about teratogens.
- Promoting the dissemination of printed information in providers' offices.
- Promoting the use of public service announcements when appropriate.
- Working with providers, AHCCCS health plans, home visiting programs, and others to target promotional efforts.
- Promoting collaboration with other resources and teams involved in health education and preventive care.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Increase in the use of folic acid by all women of childbearing age.
- Decrease in alcohol use among women of childbearing age.
- Increased awareness in population at risk of behaviors and situations associated with preventable birth defects.

- Availability of current birth defects data.
- Reduction in the rates of neural tube defects (NTD) and fetal alcohol syndrome (FAS).
- Improvement in identification, detection, and reporting of fetal alcohol syndrome.
- Increase in prenatal and preconception folate/folic acid consumption as indicated by results of the Children's
- Public awareness plan developed and implemented.
- Evaluation of public awareness efforts.
- Dissemination of folate information and fact sheet.
- Evaluation completed of current teratogen awareness programs.
- Expanded Teratogen Information Program – accomplished by funding the Arizona state legislature in 1998.

ARIZONA NEEDS:

2. PERSONS TO HAVE APPROPRIATE GENETIC RISK ASSESSMENT AND INTERVENTION FOR THE PREVENTION OF BIRTH DEFECTS AND GENETIC DISORDERS

- **Timely and appropriate referrals for genetic testing and services**
- **All pregnant women to have a prenatal risk assessment for genetic disorders/birth defects and teratogen exposure**
- **Women of childbearing age and their partners to be offered screening for genetic disorders/birth defects**
- **Genetic risk assessment to be conducted in reproductive health settings**
- **Access to an individual's genetic information to be limited**
- **All pregnant women (who are at or over the age of 35 at delivery) to be offered prenatal diagnosis for the detection of chromosomal abnormalities**

In Arizona, many people do not have access to preconception genetic screening services. Furthermore, many insurers do not cover preconception genetic testing services, because they are not considered to be “medically necessary”. Genetic testing and screening services may be more commonly considered by the provider community to have greater value after pregnancy has occurred, or after a woman has already entered prenatal care.

Arizona can do more to promote use of preconception and prenatal risk assessments that address genetic disorders. Most obstetrical care providers conduct some type of prenatal risk assessment for pregnant women. Hollister and ACOG risk assessment tools are most commonly used for this purpose. However, we do not know what percentage of providers use the tools routinely and consistently, and it is not known whether obstetrical and family practice providers have developed other additional assessments or programs related to prenatal genetic risk screening.

Accessing prenatal care early in pregnancy may not be early enough. It is thought that the most opportune time for prevention is in the preconception period when a woman has the opportunity to alter and adjust behaviors and practices to minimize teratogen exposure and other threats to early embryonic development. Clinics and health plans which provide reproductive health, well-woman care and preventive services have the opportunity to access women of childbearing age prior to conception. It is possible to conduct age appropriate risk assessments that have a genetic screening component, as well as provide screening information about teratogen usage.

Many managed care plans in Arizona are developing, or have developed, health screening and risk assessment components that focus on preventive screening services for women of childbearing age, and adult health. The health screening and risk assessment should consider ethnicity, age, and family history. These areas can be more clearly identified, coordinated, and supported throughout the state, so that genetic risk screening can be a more consistent practice among all providers. The ultimate goal of this process is to develop a framework of standardized guidelines for providers to utilize in conducting broad-based genetic risk assessment activities.

RECOMMENDATIONS AND PLANNING OPTIONS:

The following recommendations focus on promoting the utilization of established guidelines and standards for genetic health assessment and risk screening. In utilizing these guidelines, or developing our own, it will be important for Arizona policymakers to monitor national trends, as well as Arizona's own provider network regarding preconception and prenatal risk assessment services.

- **Survey providers to determine risk assessment tools currently in use, and assess the utilization of genetic screening and testing services in reproductive health/family planning networks.**

We recommend collaboration among public health professionals and health insurers to learn more about the incidence of prenatal genetic risk assessment. This combined effort could help Arizona obtain additional information about prenatal and preconception risk assessment activities. We already know that many managed care plans are focusing on health promotion and preventive health care services. AHCCCS health plans (Medicaid managed care plans) require their obstetrical providers to conduct prenatal risk assessment activities; however, there do not appear to be coordinated and consistent statewide parameters for assessing genetic risk among women of childbearing age.

There are also several networks and systems in Arizona for providing reproductive health/family planning services. Thus, it is difficult to piece together the entire spectrum of reproductive health services. Family practice and obstetrical providers, may have contracts with both commercial and AHCCCS health plans. In addition, reproductive health/family planning services are provided through family planning clinics with county and federal funds.

- **Assemble practice guidelines from other states regarding preconception, prenatal, and infertility testing.**

Arizona can develop additional documentation about what other states and regions are considering with respect to preconception and prenatal genetic risk assessment.

- **Consider developing a definition of "genetic risk assessment", and define the minimum elements of a genetics risk assessment.**

This work is fundamental to developing the framework to support genetic risk assessment guidelines for Arizona.

- **Develop a plan to promote inclusion of a statewide genetic screening component into aspects of well woman care and preventive health services for women, children, and families.**

Mountain States Regional Genetic Services Network (MSRGSN) has developed a prenatal screening tool to assess genetic issues and teratogen exposure. It is possible to adapt this tool for preconception use as well. ADHS and the genetics community can work with insurers and health care providers to consider incorporating an enhanced genetic screening component to existing developmental screening tools (e.g., AHCCCS EPSDT [teenagers], prenatal and family practice guidelines, etc.). In completing this plan, it will be critical to explore options for the development of a statewide standardized genetic risk assessment and screening tool, or, at a minimum, inclusion of a “critical data set” of information regarding genetic and teratogen screening in all preconception and prenatal risk screening tools.

The Reproductive Health Unit of ADHS (and family planning providers) should consider opportunities to review and identify genetic risk assessment attributes for possible addition to the family planning form (FHAMIS and non-FHAMIS). As a part of this effort, agencies, contractors and other community groups can collaborate in the development of a genetic risk assessment tool for non-pregnant women who access reproductive health centers and clinics.

- **Identify existing programs and agencies for collaborative links.**

A partial list of potential participating agencies and entities is included on page V-2 and V-3. Agency representatives can work together to plan, implement, and evaluate strategies for improving genetic services.

- **Develop and strengthen collaborative links with agencies and programs that are monitoring early entry into prenatal care.**

Health insurers have a vested interest in seeing that women get into prenatal care early. All AHCCCS Health plans monitor entry to prenatal care, and have developed outreach programs for early identification of pregnant women, and assessment of women who are at risk for developing complications during their pregnancy. More information is needed about how commercial insurers approach outreach activities to monitor and increase early entry to prenatal care.

- **Develop a plan with other agencies and cytogenetic testing providers to assess actual utilization of prenatal chromosomal diagnosis in women 35 years of age and older (those 35 and older who were offered testing and obtained testing, vs offered testing and declined testing.)**

This recommendation involves working with AHCCCS (Medicaid), other insurance programs, and quality improvement programs (National Committee for Quality Assurance's (NCQA) Health Employer Data Information Set [HEDIS] users) to assess incidence of those at or over age 35 at delivery who were offered (and/or utilized) services to diagnose for chromosomal abnormalities (via OB audit, or other medical record review, provider records, or encounter data information). Based on information results from the utilization study, a plan could be developed to educate the provider community about offering these services, if indicated.

- **Monitor the Arizona network for genetic counseling services.**

ADHS, health care providers, and insurers can proactively monitor the ongoing and anticipated need for genetic counseling services throughout Arizona's communities. An expected result of the screening is an increase in requests for genetic testing and counseling services. Lack of available counseling services, once a potential need is identified, is a barrier to genetic care. Lack of appointment availability, or long office waiting times can adversely impact utilization of health services. Balancing the requests for genetic counseling and services with the availability of services will be important to monitor on an ongoing basis.

- **Promote appropriate genetic risk assessment activities.**

Geneticists in Arizona can collaborate with primary care and specialty providers to develop or adopt recommendations regarding guidelines or criteria for referrals for genetic testing services. Any recommendations would be consistent with approved medical practices or other industry standards. These recommendations might include, that providers:

- Offer chromosome testing to couples who have had three or more miscarriages.
- Offer testing for persons at increased risk for genetic conditions due to ethnic background.
- Conduct more vigorous screening of prenatal clients for possible personal history of PKU, or closely follow PKU patients of reproductive age through metabolic management clinics.

- **Promote early identification of the need for genetic testing and services through provider education and community awareness.**

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Standardized or "coordinated" genetic screening evaluation tools are prepared and disseminated.
- Provider and patient surveys indicate increases in percentage of preconception and prenatal screening offered.
- Random chart audits that indicate:
 - offering of genetic counseling to pregnant women age 35 (at delivery) and older
 - offering of genetic screening to women age 35 (at delivery) and older
 - follow up was initiated when a risk was identified
 - all pregnant women have appropriate genetic risk assessments completed.

- Reproductive health clinics offer enhanced genetic risk screening component and teratogen screening.
- Completed genetic and teratogen screening plan for Arizona.
- Reproductive health forms include appropriate genetic risk assessment factors.
- Data on genetic risk assessment is returned on FHAMIS (ADHS Reproductive Health/Family Planning Management Information System) forms.
- Risk assessment survey is completed.
- Definitions of genetic risk assessment are prepared (for preconception, prenatal, pediatric, and adult), along with a data dictionary.
- A provider education plan is completed and implemented.
- A plan for developing community awareness about genetic services is completed and implemented.
- Referrals to genetic services are appropriate, based on medical chart audit.
- Referrals to genetic services are timely, based on medical chart audit.
- Referrals to services are increased in number and appropriateness.

ARIZONA NEEDS:

3. ALL INDIVIDUALS WITH A FAMILY HISTORY OF A GENETIC DISORDER TO BE OFFERED GENETIC COUNSELING AND EVALUATION

Very little is currently known about the numbers of persons in Arizona that are offered genetic counseling and evaluation because they have a family history of a genetic disorder. Those who are of child-bearing age, or who have a family history of a genetic disorder may want to know the risks for the recurrence of a genetic disorder in themselves, their offspring, or in other family members.

RECOMMENDATIONS AND PLANNING OPTIONS:

Promote insurance company awareness of the health benefits that can accrue to their members by promoting preventive genetic services.

Through networking, collaboration and coalition-building activities, ADHS and the Genetic Services Advisory Committee can act as an information resource to other agencies and health insurers throughout Arizona, and help promote understanding of preventive benefits of genetic screening and testing services. An integral step to promoting this awareness is developing additional documentation and detail regarding the national and statewide incidence of certain types of genetic disorders, and options for primary, secondary, and tertiary prevention efforts.

- **Develop a training plan for Arizona health care providers regarding genetic services and referrals.**

Providers may include a variety of target groups: insurance companies, specialty physicians, primary care providers, legislators, nurses and ancillary care providers. In addition, it may be desirable to develop a training plan or community awareness plan for other professionals or community members who may offer referrals to genetic evaluation services.

- **Convene a committee or task force to develop additional information and data about the incidence of persons in Arizona who are offered genetic counseling and evaluation due to a family history of a genetic disorder.**

Based on the results of the additional information, the committee could develop action steps and recommendations to increase the incidence and/or appropriateness of referrals to these services, when indicated.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Training plans are completed and implemented.
- Distribution of genetic screening templates to providers.
- Use of genetic screening templates by providers.

ARIZONA NEEDS:

4. GENETIC SPECIALISTS THAT ARE AVAILABLE, ACCESSIBLE, AND GEOGRAPHICALLY APPROPRIATE TO THE AT- RISK POPULATION

Many of Arizona's rural counties and communities are geographically isolated from health care services. Genetics providers conduct screening clinics in a variety of rural and urban communities on a routine basis; however, the no-show rates to appointments are high in certain areas. More work can be done to identify strategies for improving access to care for all who seek genetic counseling, evaluation, or testing services.

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Involve community and tribal leaders in designing education and information programs that are community-based, culturally appropriate, and accessible to the rural and tribal communities.**

- **Enhance the genetic services network, and improve availability and accessibility of services.**
- **Continue to strengthen links between geneticists and other health care providers, and incorporate other related agencies and teams to the network.**
- **Use the task force to identify options for improving network availability and accessibility.**
- **Continue promotion of genetics consultation contracts through ADHS.**
- **Work with genetics community, providers, consumer groups, and CRS to develop information for Arizona health care providers and consumers on the availability of genetic clinics, and the names and addresses of all genetic service providers in Arizona.**
- **Develop a plan to refine and clarify the availability and accessibility standards for genetics services in Arizona.**

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Completed information for providers and community on available genetics network and number of patients seen.
- Comparison of expected presentation in genetics clinics, given rate of birth, with the number of patients seen with a genetic disorder.
- Increase in numbers of genetics referrals and numbers of persons accessing genetic services.
- Reduction in no-show rates to genetics clinics.
- Increase in the number of pregnant women accessing genetic services.
- Options for improving availability and accessibility developed by a Task Force.

ARIZONA NEEDS:

- 5. GENETIC DIAGNOSTIC PROCEDURES AND GENETIC TESTS TO BE PERFORMED FOLLOWING COMPREHENSIVE GENETIC EVALUATION BY APPROPRIATELY TRAINED SPECIALISTS.** We recommend that genetic diagnostic and testing procedures be conducted only after a comprehensive genetic evaluation. The genetic evaluation and test procedures would be performed only by appropriately trained specialists who have met educational and/or training requirements in the technical, diagnostic, and interpretive aspects of the procedures.

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Prepare a document outlining what consumers should look for in seeking genetic counselors and genetic testing services.**

Consumers need relevant information about available choices and options for genetic testing services. This document can be a simple, user-friendly reference to help health care consumers and the general public identify important factors to consider in seeking services. For example, it is helpful for consumers to know that all prenatal diagnostic testing should include a pretest counseling session, informed consent, appropriate screening or diagnostic tests, and follow-up counseling session. The availability of this document can be invaluable in assisting and empowering Arizonans to make informed choices about these services.

- **Prepare and disseminate lists of resources available to all providers regarding genetic testing.**

These resources can help educate providers, including physicians, hospitals, laboratories, and health insurers/managed care health plans, about genetic service providers in Arizona. The document would list Arizona's genetic service providers and explain their qualifications, and the types of diagnostic testing or counseling services that can be referred to these providers. The document can also outline what type of specialist or clinician is appropriate to perform which type of procedures, as well as appropriate standards for conducting genetic tests, or making referrals for genetic diagnostic, testing, or counseling services.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Encounter data or laboratory claims data indicates that procedures were performed by appropriately trained specialists.
- Genetics evaluation and counseling precedes the prenatal diagnostic procedures.

ARIZONA NEEDS:

6. HEALTH CARE PROVIDERS TO BE KNOWLEDGEABLE AND INFORMED ABOUT GENETIC RISKS, SERVICES AND RESOURCES

- All providers to be knowledgeable about laboratory resources and specimen submission procedures
- All providers to be informed about selected genetic risks
- All providers to be aware and informed of how to access genetic services.
- Health and social service training programs to include a genetics component

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Improve laboratory submission for newborn screening and referral for other genetic tests.**

There are many options for improving laboratory submission and testing for newborn screening disorders. Many of our recommended options focus on collecting performance related data, and training providers in appropriate submission procedures:

- Collect data regarding the number of unacceptable specimens currently submitted.
- Prepare a genetic test submission manual containing laboratory testing requirements for common genetic tests and newborn screens
- This manual could be a summary document for physicians, lab technicians, and submitters, regarding testing parameters and requirements for all types of genetic tests needed by citizens in Arizona. The manual would include labs, location, clinical indications, timing or prerequisites for tests specimen gathering and packaging requirements, labeling, addressing, and time frames, location address, and contact persons.)
- Develop and implement a training plan for educating health care providers regarding specimen submittal. Train providers in the proper submission of specimens.
- Develop and/or enhance ongoing reporting or data collection mechanisms for tracking unacceptable specimens.
- **Promote early identification and detection of need for genetic testing and services through provider education of genetic risks and community awareness.**

Several steps are recommended in this process, including:

- Collecting information about how referral processes to genetic services are initiated.
- Identifying continuing education and inservice opportunities for physician providers.
- Documenting a plan for informing providers of how to access genetic services.
- Developing professional education programs for preconception, prenatal, children, and adult issues.
- Educating medical directors on the need for primary care providers to offer their patients genetic diagnostic services.
- Educating providers about genetic risks and the referral process.
- **Collaborate with consumer groups and parent/child advocacy organizations to educate providers about the availability of family support organizations.**

There are many support groups and organizations for parents, families, or persons with genetic disorders. It is important for providers to be aware and informed of these organizations and how to make referrals. The support organizations often have links to new information on the latest trends in diagnostics and therapeutics. Recommended activities to promote the provider education process include:

- Surveying health service, social service, and medical education/training programs in Arizona to obtain information about genetics curriculum content.
- Promote inclusion of enhanced genetics component in health services/medical education curricula.
- Link with others to identify opportunities for collaboration.
- Based on information and assessment, develop a plan (with educational institutions) to target and improve areas in need of additional coverage in curricula.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Laboratories receive a higher percentage of specimens that are acceptable for testing.
- Provider education program is completed.

- Referrals to genetic services are appropriate and timely.
- The number of referrals to genetic services are increased.
- Analyses of referral processes are completed.
- GSA assessment/directory to providers is distributed.
- Provider and public survey measures of knowledge and general awareness are monitored.
- Results of Arizona curriculum assessment of genetic services (for health service, social service, and medical education providers).
- Plan of action for genetics curriculum promotion is developed.

ARIZONA NEEDS:

7. GENETIC LABORATORY SERVICES TO BE OF HIGH QUALITY.

- Genetic laboratory services that are available, accessible, timely
- Genetic laboratories that have appropriate quality control audits and/or accreditation
- Laboratory directors to be appropriately certified

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Improve availability and accessibility of laboratory services.**
 - Convene a task force or subcommittee to explore options for development and support of additional diagnostic testing capability for genetic services in Arizona.
 - Identify and document existing networks of genetic laboratory providers and facilities used by Arizona health care providers and organizations. Make this list available to provider community through the education and orientation process.
 - Evaluate the current system, including specimen transportation, testing time, cost, protocol, interpretation of results, and other issues as identified.
 - Produce an evaluation report to the Genetic Services Advisory Committee regarding laboratory services in Arizona.

- **Assure a mechanism for access to genetic laboratory testing when indicated, to ensure payment for genetic laboratory testing is available to all who require it.**
- Convene a working group of the GSAC to explore options for payment, including clinicians, finance, managed care organization representatives, and AHCCCS.
- **Promote appropriate accreditation for laboratory services that provide genetic screening and testing services for Arizona clients.**
 - Develop network and links with laboratory regulation agencies for genetic lab testing.
 - Research current requirements for laboratory certification in Arizona, and determine availability of other special certification/requirements for laboratories that handle metabolic or genetic testing specimens.
 - Promote the use of standards of practice used by cytogeneticists and consultants to laboratories that provide genetic testing services through the inclusion of practice standard requirements in existing state contracting systems (e.g. AHCCCS)

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Testing capabilities in Arizona are improved.
- Laboratories meet appropriate turnaround times for test results.
- The list of appropriately certified laboratories includes all laboratories used for genetic testing by Arizonans.
- Employee or contractor listings includes appropriately certified individuals.

ARIZONA NEEDS:

8. INCREASED PUBLIC AWARENESS OF GENETIC ISSUES AND GENETIC SERVICES

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Collaborate with providers, insurance companies, and consumers to document a plan for informing consumers on how to access genetic services.**
 - Gather information from insurance companies regarding the availability of internal genetics education programs.

- Gather information from insurance companies and the provider community about how referral processes to genetic services are initiated for preconception, prenatal, child, and adult genetic services.
- Conduct a focus group for input regarding genetic service needs.
- Review currently available consumer information.
- Develop a statewide genetic services awareness program with input from consumers and providers.
- Develop public service announcements to increase awareness of preventable genetic conditions and availability of genetic services.
- Seek support for the project through the March of Dimes, federal grants, or other funding sources.

IMPROVEMENT INDICATORS

- The following factors may be used to measure whether program improvements have been achieved:
- Plan for improving consumer/public awareness is completed.
- Consumer/public awareness plan is implemented.

Measures of knowledge and general awareness by providers and public are surveyed.

ARIZONA NEEDS:

9. **ALL INDIVIDUALS ARE INFORMED OF, AND THOROUGHLY COUNSELED AS TO THE BENEFITS, RISKS, AND LIMITATIONS OF GENETIC TESTING, DIAGNOSIS AND TREATMENT, AND THE POSSIBLE CONSEQUENCES OF GENETIC TESTING.**

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Provide complete, accurate and clear consent information and other information for genetic testing.**
 - Conduct consumer, ADHS, and provider focus groups.
 - Make recommendations and suggestions regarding informed consent for genetic testing.

- **Develop and promote genetic education programs for providers that incorporate some instruction in genetic counseling.**
 - Gather materials currently available, and disperse to focus groups.
 - Review currently available information during focus group meetings.
 - Develop educational materials to facilitate the provision of this instruction.
 - Develop and implement multi-media training sessions and/or workshops to include opportunities for interactive practice in counseling about genetic services and testing.
 - Provide educational materials to providers/counselors regarding genetic testing and treatment options.

In a collaborative effort, develop appropriate information and “minimum standards regarding benefits, risks, and options for genetic testing”, to be shared with clients, parents, and providers.

- Review American Medical Association, American College of Medical Genetics, National Society of Genetic Counselors, American College of Obstetrics and Gynecology (ACOG), American Academy of Pediatrics, Alliance of Genetic Support Groups, and other guidelines for informed consent, and patient rights and responsibilities.
- Develop or refer to guidelines for altered medical management of the pregnancy in cases with altered maternal (e.g., maternal PKU) or fetal genetic conditions (e.g., Caesarean-section of pregnancy in baby with spina bifida).
- Review existing statements, and develop a statement of client rights and responsibilities related to genetic testing services.
- Provide client rights and responsibilities materials to providers, consumers, and legislators.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Analysis of counseling and informed consent procedures.
- Collected minimum standards for genetic testing information are established.
- Statement of client rights and responsibilities is developed.

- Client satisfaction surveys are developed, implemented, and evaluated.
- Medical record audits indicate that genetic counseling is provided prior to prenatal tests, such as CVS or amniocentesis (i.e., investigate opportunities to obtain information through other agencies that are conducting medical audit projects, or quality assurance/continuous quality improvement activities.)

ARIZONA NEEDS:

10. ALL PERSONS TO BE PROTECTED FROM DISCRIMINATION RESULTING FROM GENETIC TESTING, SERVICES, OR DISORDERS

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Educate the legislative community about genetic services. Prepare educational information in response to requests from various governmental agencies or legislative bodies.**
 - Education Committee assists in developing appropriate educational materials for the Arizona legislature regarding newborn screening and genetic lab testing, etc.
 - Recommend improvements in legislation when needed.
- **Promote community integration of newly developed or approved legislation regarding genetic discrimination.**
 - Monitor the implementation of new legislation.
- **Develop systems and processes to ensure that genetic information is not used to make health care and other insurance prohibitively expensive.**
 - Search for data, info on cases (through Department of Insurance (DOI) and other sources), compile case data, promote/support education on this issue.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Public information about genetic testing is incorporated into the community.
- Persons at genetics information repository do not release information to unauthorized persons.

ARIZONA NEEDS:

11. IMPROVED DATA COLLECTION AND ANALYSIS REGARDING BIRTH DEFECTS, GENETIC DISEASES, GENETIC SERVICES AND HEALTH OUTCOMES
RECOMMENDATIONS AND PLANNING OPTIONS:

- Utilize working group of GSAC to conduct an in-depth analysis of genetic services data reporting requirements.
- Develop a core set of questions and issues regarding genetic services data, such as information regarding genetic risk assessment, genetic education, and utilization reporting.
- Study genetics services data and reporting requirements from all systems: AHCCCS, state and private adoption agencies, Newborn Screening Program, CRS, specialty providers, and commercial insurers.
- Determine parameters, limitations and restrictions for access to genetic services data and information.
- Create a minimum data set of genetic services information, including a data dictionary (outlines definitions for data elements).

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Analysis of data reporting requirements is completed.
- Minimum data set is completed.
- Data dictionary is completed.
- Plan for integrating, collecting and reporting data and genetic information is developed and implemented.

ARIZONA NEEDS:

12. PERSONS TO HAVE ACCESS TO SINGLE-SITE MULTI-SPECIALTY, INTERDISCIPLINARY TEAMS WHEN APPROPRIATE FOR THE CARE AND TREATMENT OF IDENTIFIED GENETIC DISEASES.

Children have access to multi-specialty, interdisciplinary care for several types of genetic disorders through the OCSHCN/CRS Program.

Multi-specialty, interdisciplinary care for children with special health care needs considers the needs of the whole child and family, and provides continuity and consistency in the treatment of children with multiple medical or social needs. This care delivery system should also be evaluated for adult genetic services.

- **Adult Services**

- Obtain more information about adult multi-specialty, interdisciplinary clinics.
- Develop a task force to examine multi-specialty, interdisciplinary care options for genetic disorders. Define the task force role, and develop other venues and/or options for multi-specialty, interdisciplinary care for adults.
- Identify barriers for adults to access multi-specialty, interdisciplinary care and services for genetic disorders (e.g., geography, personnel, cost, space and transportation).
- Consider network expansion options, such as case management models, etc. Other payer and network relationships need to be identified, including use of children's networks for care of adults.

- **Pediatric Services**

- Obtain more information about pediatric multi-specialty, interdisciplinary clinics.
- Develop a task force to examine multi-specialty, interdisciplinary care options for genetic disorders. Define the task force role, and develop other venues and/or options for multi-specialty, interdisciplinary care for children.
- Identify barriers for children to access multi-specialty, interdisciplinary care and services for genetic disorders (e.g., geography, personnel, cost, space and transportation).
- Consider network expansion options, such as case management models, etc. Other payer and network relationships need to be identified.

- **Implementation**

- Explore options with CRS for expanding medical eligibility and funding for CRS services, by diagnosis first, then expand for age after diagnosis.
- Develop a plan to improve multi-specialty, interdisciplinary services and clinics for preconception, prenatal, children and adult services.
- Implement the plan to improve multi-specialty, interdisciplinary services and clinics for preconception, prenatal, children and adult services.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Completed plan for addressing multi-specialty, interdisciplinary service options in Arizona.
- Compare number of spina bifida births with the number of spina bifida patients seen in clinics.

ARIZONA NEEDS:

13. ALL ADOPTIVE CHILDREN TO HAVE ACCESS TO GENETIC INFORMATION ABOUT THEIR BIOLOGIC FAMILY

- All birth parents placing a child for adoption to be offered genetic screening.

RECOMMENDATIONS AND PLANNING OPTIONS:

- Review current legal and legislative requirements for genetic information gathering for adoptions (public and private).
- Review national standards or guidelines from other states regarding collection of genetic information prior to adoption.
- Identify possible resources that could be used to assist birth parents to provide complete genetic information (e.g., questionnaire, family history taken).
- Develop and implement an educational program with adoption agencies regarding the importance of genetic information.
- Work with state and private adoption agencies to implement options for improving non-identifying data collection of genetic information (family history or actual results of genetic testing and diagnosis from birth parents to be provided to adoptive parents).
- Evaluate if birth parents are supplying genetic information and if it is available to the adoptive child, and/or adoptive parents.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Adoption information form contains additional questions on genetic history (genetic risk assessment).
- Genetics education program is implemented.

- Completed tally sheets of persons requesting and receiving genetic information.

ARIZONA NEEDS:

14. COMPLETE GENETIC BACKGROUND INFORMATION TO BE COLLECTED ABOUT EGG AND SPERM DONORS

RECOMMENDATIONS AND PLANNING OPTIONS:

- *Review current legal and legislative requirements, and national standards and guidelines for genetic information gathering for egg and sperm donation.*
- *Identify possible resources that could be used to assist donors in providing complete information (e.g. questionnaire, family history taken), that would be available to egg and sperm recipients.*
- *Work with state and private adoption agencies, infertility clinics, and/or assisted reproduction facilities to investigate options for improving data collection of genetic information of birth parents.*
- *Develop and implement an educational program, including a conference for infertility/OB providers, sperm bank staff.*
- *Implement donor carrier testing for common genetic disorders when available and provide appropriate follow-up.*
- *Evaluate whether donors provide information, receive carrier testing and follow-up information.*
- *Evaluate surrogate mothers for metabolic disorders which might affect the fetal environment.*

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Completed conference/seminar sessions.
- Sperm and egg specimens accompanied by comprehensive genetic information.

ARIZONA NEEDS:

15. **ALL PERSONS TO EXERCISE FREE CHOICE WITH RESPECT TO GENETIC TESTING OR TREATMENT IDENTIFIED AS A CORE VALUE FOR THE SYSTEM AND PLAN. THE GENETIC SERVICES ADVISORY COMMITTEE COORDINATES ALL ACTIONS AND PROJECTS TO SUPPORT CORE VALUES. TARGET COMPLETION IS ONGOING.**

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Ensure that use of genetic testing services is voluntary.**
 - **Conduct a survey of providers who perform genetic tests. Include genetic counselors in the survey.**
 - **Assess processes, procedures, and documentation produced and provided during pretest genetic counseling.**
 - **Investigate processes and procedures for providing post-procedure interpretation and counseling, and guidelines for disclosure of test results and information.**
 - **Promote use of non-directive counseling.**
 - **Develop an informed consent checklist (using input and review from geneticists and consumers) for counselors and providers to use in discussing genetics testing, treatment, and options with clients and families.**
 - **Include education themes in curricula, health services education, medical continuing education, consumer/prenatal education, provider training, patient Bill of Rights, and information about procedures to lay health workers.**
 - **Incorporate into training at all levels--information about: 1) non-genetic trained individuals providing services, and 2) processes for promoting the concept of voluntary services.**
- Gather policy statements related to presymptomatic genetic testing in children.**

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Client satisfaction survey indicates that persons receiving genetic services did so voluntarily.
- Consumer forum(s) results indicate that services are voluntary.

ARIZONA NEEDS:

17. GENETIC SERVICES TO BE FAMILY CENTERED, CULTURALLY COMPETENT, INTEGRATED, GEOGRAPHICALLY APPROPRIATE, AND FOCUSED ON THE MAINTENANCE OF HEALTH

IDENTIFIED AS A CORE VALUE FOR THE SYSTEM AND PLAN. THE GENETIC SERVICES ADVISORY COMMITTEE COORDINATES ACTIONS AND PROJECTS TO SUPPORT CORE VALUES. TARGET COMPLETION IS ONGOING.

RECOMMENDATIONS AND PLANNING OPTIONS:

- **Establish a multi-disciplinary task force/subcommittee to identify strategies for improving coordination of care and services for all persons who need or receive genetic services.**
- **Investigate current case management networks existing in Arizona for care and treatment of persons affected with genetic disorders.**
- **Enhance communication and referral networks between rural primary care providers and urban based geneticists, multi-disciplinary treatment specialists and insurance payors.**
- **Explore the possibility of increasing access to information for the public and consumers, through technology such as Medline, Internet, or the ADHS Websites.**
- **Continue to strengthen links with other OWCH teams, home visiting programs, and parent support groups.**
- **Explore opportunities to expand state funding through OCSHCN/CRS for “linkage” genetic testing for families in cases where definitive diagnosis for a child requires that family members be tested. Long range plans should include exploration of options to offer additional population based genetic susceptibility screening, both for children and adults, including options for insurance coverage for families of affected children.**
- **Work with providers and parent support groups to develop and implement a confidential satisfaction survey for persons who receive genetic screening or testing services.**
- **Add questions on genetic services to the next OCSHCN survey.**
- **Use results of the satisfaction survey to develop a plan for improvement.**
- **Work with wellness and prevention programs to identify strategies for wellness promotion in persons with genetic conditions.**

- Promote expansion of the family advocate and support network for service coordination resources.
- Identify and link with current follow up and coordination programs, and encourage new programs where there are gaps in service.
- Explore linking with Office of Minority Health.
- Prepare a Spanish translation for all written information provided to the public regarding genetic services.
- Train all professionals, including medical providers, hospital personnel, and teachers, in cultural awareness and sensitivity issues surrounding genetic services.
- Promote the availability of verbal translation services and service coordination to meet the needs of individuals seeking genetic services.
- Consider availability of translation services for tribal populations, and appropriateness of information in written or spoken languages.
- Promote integration and coordination of genetic services with other aspects of primary care.
- Promote prevention and health maintenance attributes of genetic services.
- Develop sensitivity training programs for providers that support holistic treatment of children and families, incorporating the principles of family centered care.
- Involve consumer advocacy groups, health plan/insurance member service representatives, and other family advocacy agencies in efforts.
- Involve parent support groups and consumer advocates in designing materials for the genetics program and the public.
- Develop documentation for the public that is user-friendly and easy to read.
- Promote education in all sectors of the population regarding genetic contribution to common diseases as well as rare hereditary disorders.
- Involve community and tribal leaders in designing education and information programs that are community-based, and accessible to the rural and tribal communities.

IMPROVEMENT INDICATORS

The following factors may be used to measure whether program improvements have been achieved:

- Task force is organized.
- Provider education programs include training in appropriate communication with families.
- Results of clinic patient survey indicate satisfaction.
- At-risk population brochures are developed and distributed.
- Description of Genetic Service care system is produced and disseminated.
- Education/training packages for health care providers/trainers are completed.
- Primary care provider's medical records make reference to, and contain information about, genetic screening and testing services provided to their patients.
- Written materials are available in English/Spanish or predominant second language.
- Materials are written at an appropriate reading level.
- Results of satisfaction surveys indicate that consumers are satisfied with services.
- Training programs have been completed and evaluated.

APPENDIX A

ORGANIZATION CHART

ARIZONA DEPARTMENT OF HEALTH SERVICES OFFICE OF WOMEN'S AND CHILDREN'S HEALTH

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APPENDIX B

GLOSSARY

APPENDIX B GLOSSARY

ABGC

American Board of Genetic Counseling.

ABMG

American Board of Medical Genetics. A member board of the American Board of Medical Specialties. Certifies medically qualified practitioners of the genetic subspecialties..

ACMG

American College of Medical Genetics

ACOG

American College of Obstetrics and Gynecology.

ADHS

Arizona Department of Health Services.

AFP

Alpha fetoprotein, also known as the single marker screen when referring to maternal serum screening. A fetal protein that can be used to screen pregnant women for potentially at-risk pregnancies.

AFP2

Alpha fetoprotein and human chorionic gonadotropin (HCG), also known as the double marker screen when referring to maternal serum screening.

AFP3

Alpha fetoprotein, human chorionic gonadotropin (HCG) and estriol, also known as the triple marker screen when referring to maternal serum screening.

AFAFP

AFP collected from amniotic fluid (AFAFP). May confirm or refute MSAFP result.

ALLELE

The actual nucleotide sequence of genes on the chromosome. Changes in sequence from one allele to another are the result of mutations, and these mutations can be transmitted to subsequent generation.

AMNIOCENTESIS The removal of a small amount of fluid from a pregnant woman's uterus with a thin needle, usually done to determine the status of the fetus in the second or third trimester. Also used to monitor Rh isoimmunization and maturity of the fetus.

ANENCEPHALY

A congenital birth defect caused by the improper closure of the neural tube at the anterior end resulting in absence of the skull and partial or complete absence of the cerebral hemispheres.

ASHG

American Society of Human Genetics.

ASSESSMENT

The systematic process of collecting relevant client data for the purpose of determining actual or potential health problems and functional status. Methods used to obtain data include interviews, observations, physical examinations, review of records, collaboration with colleagues, and consideration of applicable literature and research.

AUTOSOMAL

Determined by a gene on one of the chromosomes other than a sex chromosome (X or Y).

BIRTH DEFECT

Abnormal congenital condition ranging from minor to severe that may result in debilitating disease, a physical or mental disability, or early death. Birth defects may or may not have a genetic cause.

CAP

College of American Pathologists. Oversees accreditation of medical laboratories, including genetics laboratories.

CDC

Center for Disease Control and Prevention.

CFHS

Community and Family Health Services (in Arizona Department of Health Services).

CERTIFICATION

The formal process by which clinical competence is validated in a speciality area of practice, typically involving examination of the applicant's knowledge.

CLIA

Clinical Laboratory Improvement Act.

CLIENT

1) A person with or a couple who have had a child with a genetic condition; 2) a presymptomatic person or family at risk for a genetic condition; 3) a person who is susceptible to a disease which has a genetic component; 4) a person or couple who are at risk for having a child with a genetic condition; 5) a person or group who needs or requests genetic information; or 6) a group, community, or population with or at risk for genetic condition

CLINICAL GENETIC SERVICES

Services designed to screen, diagnose, treat, or identify risks for disorders or conditions with a genetic component. Clinical genetic services also includes counseling and support services to assist clients in making voluntary decisions regarding testing and/or treatment.

CORN

Council of Regional Networks for Genetics Services.

CRS

Children's Rehabilitative Services.

CHILDBEARING AGE

Women who are age 15 to 44 years (Maternal Child Health and Center for Disease Control definitions).

CONGENITAL

Present at birth. Congenital anomalies may or may not be genetic.

CONSANGUINITY

Relationship by descent from a common ancestor. A union between two persons who are 1st or 2nd degree relatives are at a increased risk for having offspring with a recessive genetic condition or multifactorial disorder than an unrelated couple.

CVS

Chorionic villus sampling. A prenatal test used to detect some types of genetic conditions. CVS sampling may be done between 10-12 weeks of gestational age.

DOMINANT

A characteristic or disorder expressed in the heterozygote (offspring that inherits two different alleles (one from each parent) at a gene locus). The dominant gene is the one that is expressed, when the two genes of any gene pair are different alleles, and only one of those alleles is expressed. A condition is dominant if only one of the 2 gene copies inherited from the 2 parents (one copy each) is needed to transmit the condition to the offspring. The risk of transmitting a disorder determined by a dominant gene is 50%.

DOUBLE MARKER SCREEN

Alpha fetoprotein (AFP) and human chorionic gonadotropin (HCG).

DYSMORPHOLOGY

The study of abnormal physical development, e.g., as might be found in a syndrome of genetic or environmental etiology.

FAS

Fetal alcohol syndrome.

Fetal alcohol effects.

FISH

Fluorescent in situ hybridization.

FOLIC ACID

A member of the B complex vitamins, found naturally in green plants, fresh fruit, liver, and yeast. Consumption of adequate amounts of folic acid prior to and during pregnancy can reduce the risks of neural tube defects in infants.

FOLATE

The food source for obtaining folic acid.

IHS

Indian Health Services.

GENETIC CONDITION

A disease which is caused or influenced by genes and may or may not be transmitted from parent to offspring; sometimes referred to as a genetic disorder or genetic disease.

GENETIC DIAGNOSIS

Cytogenetic, biochemical, or molecular studies, and/or identification of a clinical phenotype that identifies the individual as having a genetic condition.

GENETIC SCREENING

Testing that refines the calculation of an individual's risk of manifesting or transmitting a genetic condition or having offspring with a birth defect. Types of genetic screening include neonatal screening, prenatal screening, and population screening. Effective screening programs meet specific criteria; 1) the genetic condition is relatively frequent in a population; 2) the test is highly sensitive, specific and relatively inexpensive; 3) the benefits of the program outweigh its psychological, social, ethical, economical "costs"; 4) the screening test results can be confirmed by the diagnostic tests in a timely manner; 5) treatment and/or reproductive options are available for individuals testing positive; and 6) there are appropriate counseling and support services for the person(s) identified as being at risk for manifesting or transmitting genetic conditions or having offspring with a birth defect.

GENETIC THERAPEUTIC MODALITIES

Treatment for a genetic disorder. Some of these approaches include but are not limited to: 1) dietary modification, e.g., for phenylketonuria and familial hypercholesterolemia; 2) replacement of defective gene (gene therapy), e.g., for inherited immune deficiencies; 3) replacement of deficient enzyme, e.g., for Gaucher disease; 4) other gene product replacement, e.g., as in hemophilia (factor VII, IX); 5) other medical therapies, e.g., penicillamine for Wilson disease or allopurinol for hyperuricemia; and 6) surgical approaches, e.g., renal transplantation for polycystic kidney disease.

HEMOGLOBINOPATHY

Hemoglobinopathy is one of the seven disorders screened by the Newborn Screening Program. Hemoglobin is a complex protein within red blood cells that is responsible for carrying and releasing oxygen to body tissues. The term hemoglobinopathy refers to the large family of inherited genetic disorders of hemoglobin. These disorders fall into three major types: structural variations in the hemoglobin; thalassemias (reduced rates of synthesis of proteins that comprise hemoglobin); and hereditary persistence of fetal hemoglobin. Hemoglobinopathy represents the single most common genetic disorder in the human population. Sickle cell anemia is caused by a variation in the structure of hemoglobin.

HUMAN CHORIONIC GONADOTROPIN (HCG)

A hormone synthesized by the placenta that supports the pregnancy. The presence of HCG in a woman's urine suggests pregnancy.

HOMOCYSTINURIA

One of the seven disorders screened by the Newborn Screening Program. This inherited disorder is caused by a decrease in the activity of a certain enzyme, which causes the buildup of excessive amounts of homocystine and methionine. There are no clinical symptoms of this disorder in the newborn period, however, children may later develop seizure disorders, neurological damage, cataracts and dislocated lenses, osteoporosis and connective tissue damage. Treatment may be in the form of high doses of B6 and/or dietary restriction of methionine. Not all forms are responsive to B6. The newborn screening test may be done too early to identify all children with homocystinuria, therefore, some states have instituted the requirement of a second screen.

MOM

Multiples of the median, method by which alpha fetoprotein test results are reported

MSAFP

Maternal serum alpha fetoprotein

MSRGSN

Mountain States Regional Genetics Services Network, one of ten regional networks in the Council of Regional Networks for Genetic Services (CORN). Six states included in the MSRGSN are Arizona, Colorado, Montana, New Mexico, Utah, and Wyoming.

NTD

Neural tube defect, caused by failure in closure of the neural tube, which may result in disorders such as spina bifida, anencephaly, encephalocele

OCSHCN

Office of Children with Special Health Care Needs in Arizona Department of Health Services

OWCH

Office of Women's and Children's Health in Arizona Department of Health Services.

PCP

Primary care provider.

PEDIGREE

In medical genetics, a diagrammatic representation of a family history, indicating individuals affected with or at risk for a genetic condition and their relationship to the client.

PHENYLKETONURIA

PKU; one of the seven disorders screened by the Newborn Screening Program. This metabolic disorder is an inherited defect in which the body cannot use the amino acid phenylalanine properly. Phenylalanine is found in all dietary protein. Excess levels of phenylalanine metabolites accumulate in the blood, and can cause severe brain damage and retardation if left untreated.

PREGNANCY RATE

The sum of all live births, fetal deaths, and abortions (during a time period, per 1,000 population) of women 15-44 years old.

PRESYMPTOMATIC

Inherited gene alteration known to cause an adult onset condition. That is genetically identified before the appearance of symptoms. Example: the altered gene for Huntington disease can be detected through molecular testing before symptoms appear.

PUBS

Percutaneous umbilical blood sampling. A sampling of the fetal blood.

RECESSIVE

A characteristic or condition only expressed when both alleles at a genetic locus are altered; a trait or gene that is expressed only if the individual is homozygous for a given allele (i.e., has two copies of the allele that are alike, one from each parent). For example, the gene for blue eye color is homozygous. In order to have blue eyes expressed as a trait, offspring must inherit a blue allele from each parent.

SICKLE CELL DISEASE

An inherited disease of the blood, affecting hemoglobin. Sickle cell disease is characterized by changes in the biochemical and physical properties of red blood

cells, resulting in the “sickle” shape. Sickle cell disease occurs when an individual inherits a sickle cell gene from each parent.

SICKLE CELL TRAIT

In persons with a sickle cell trait, one gene for sickle cell is inherited from one parent, and a normal gene is inherited from the other parent. Persons with sickle cell trait usually do not manifest symptoms of sickle cell disease.

SUSCEPTIBLE

The presence of an altered gene or genes identified through molecular DNA techniques that increase an individual's risk for developing a particular disorder or disease. The presence of a gene or genes does not assure that an individual will become affected, but places them at increased risk. Example: some common multifactorial conditions such as diabetes mellitus and some cancers.

TERATOGEN

A physical or chemical agent that is associated with an increased risk of birth defects. Alcohol is a teratogen known to cause fetal alcohol syndrome.

TRIPLE MARKER SCREEN

Alpha fetoprotein (AFP) and human chorionic gonadotropin (HCG) and estriol.

X-LINKED PATTERN OF INHERITANCE

Pattern of inheritance for genes located on the X chromosome, often called sex-linked inheritance. An X linked recessive conditions usually affect more males than females, because females have the protection of their second X chromosome. X-linked dominant conditions require only one of the X chromosomes to have a gene mutation in order to cause disease onset.

APPENDIX C

REFERENCES AND RESOURCES

APPENDIX C REFERENCES AND RESOURCES

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APPENDIX D

OFFICE OF WOMEN'S AND CHILDREN'S HEALTH PROGRAM DESCRIPTIONS

Note:

Current program descriptions for Women's and Children's Health can be found on the OWCH web site at: <http://www.hs.state.az.us/phs/owch>

APPENDIX E

ACRONYMS

ACRONYMS

AAHC	Arizona Adolescent Health Coalition
AAP	American Academy of Pediatrics
ABA	American Bar Association
ACOG	American College of Obstetricians and Gynecologists
ADE	Arizona Department of Education
ADHS	Arizona Department of Health Services
AHCCCS	Arizona Health Care Cost Containment System
APHA	American Public Health Association
APT	Arizona Perinatal Trust
A.R.S.	Arizona Revised Statutes
ASCAP	Arizona Sickle Cell Anemia Program
ASDB	Arizona State School for the Deaf and Blind
ASU	Arizona State University
AWHS	Arizona Women's Health Survey
AzEIP	Arizona Early Intervention Program
BRFSS	Behavioral Risk Factor Surveillance Survey
C.A.R.E.	Children's Assistance Resource Event
CATS	Client Automated Tracking System (Formerly called FHAMIS)
CDA	Child Development Associate
CDC	Center for Disease Control and Prevention
CFHS	Community and Family Health Services
CH	Congenital Hypothyroidism
CHD	County Health Department
CHN	Community Health Nurse
CIC	Children's Information Center
CISS	Community Integrated Service Systems
CRS	Children's Rehabilitative Services
DDD	Division of Developmental Disabilities (DES/DDD)
DES	Department of Economic Security
EPSDT	Early Periodic Screening, Diagnosis and Treatment (Services)
FHAMIS	Family Health Automated Management Information System
FP	Family Planning
FY	Fiscal Year
HCG	Human Chorionic Gonadotropin
HIV/AIDS	Human Immunodeficiency Virus (Infection)/Acquired Immunodeficiency Syndrome
IHS	Indian Health Service
LBW	Low Birth Weight
MCH	Maternal and Child Health
MSRGSN	Mountain States Regional Genetic Services Network
MSAFP	Maternal Serum Alpha Fetoprotein
NICP	Newborn Intensive Care Program
NICU	Newborn Intensive Care Unit

OAD	Office of Assistant Director
OB/GYN	Obstetrics and Gynecology
OCSHCN	Office for Children with Special Health Care Needs
ONS	Office of Nutrition Services
OOH	Office of Oral Health
OWCH	Office of Women's and Children's Health
PKU	Phenylketonuria
PUBS	Percutaneous Umbilical Blood Sampling
S.O.B.R.A.	Sixth Omnibus Budget Reconciliation Act
SHRI	School Health Risk Inventory
SIDS	Sudden Infant Death Syndrome
SPRANS	Special Projects of Regional and National Significance (Title V Funds)
SSA	Social Security Administration
SSI	Supplemental Security Income
STD	Sexually Transmitted Disease
USDA	U. S. Department of Agriculture
VLBW	Very Low Birth Weight
VS	Arizona Vital Statistics
WIC	Special Supplemental Food Program for Women, Infants and Children

APPENDIX F

ARIZONA LAWS AND RULES REGARDING GENETIC SERVICES

RULES

CHAPTER 14. DEPARTMENT OF HEALTH SERVICES

LABORATORIES

ARTICLE 5. TESTS FOR METABOLIC DISORDERS AND HEMOGLOBINOPATHIES

Section

R9-14-501. Definitions

R9-14-502. Testing of Newborns

R9-14-503. Persons and Health Care Facilities Responsible for Tests

R9-14-504 Parent or Guardian Education

R9-14-505. Collection of Screening Fees

ARTICLE 5. TESTS FOR METABOLIC DISORDERS AND HEMOGLOBINOPATHIES

R9-14-501. Definitions

In this Article, unless context otherwise requires:

- 1. "Biotinidase deficiency" means a congenital metabolic disorder characterized by abnormal biotinidase production which causes mental retardation if not treated early in life.**
- 2. "Committee" means the newborn screening program committee appointed by the Director.**
- 3. "Congenital hypothyroidism" means a metabolic disorder characterized by a deficiency of thyroid hormone (thyroxin) production which causes mental and physical retardation if not treated early in life.**
- 4. "Department" means the Department of Health Services.**
- 5. "Director" means the Director of the Department of Health Services.**
- 6. "Galactosemia" means a congenital metabolic disorder characterized by abnormal galactose metabolism which causes mental retardation or death if not treated early in life.**
- 7. "Health care facility" means any establishment, public or private, that provides facilities for obstetrical care and care to a newborn.**
- 8. "Health care provider" means the physician, nurse practitioner, or licensed midwife caring for the newborn after delivery.**

9. "Hemoglobinopathies" mean a group of inherited diseases characterized by an abnormality in the production and function of hemoglobin.

10. "Homocystinuria" means a congenital metabolic disorder characterized by abnormal methionine metabolism which causes mental retardation if not treated early in life.

11. "Initial screen" means laboratory procedures performed on the first acceptable specimen of blood to detect the presence of metabolic disorder and/or a hemoglobinopathy.

12. "Maple syrup urine disease" or "M.S.U.D." means a congenital metabolic disorder of branch chain amino acid metabolism which causes mental retardation or death if not treated early in life.

13. "Newborn" means an infant 30 days of age and under for whom a certificate of live birth is required by A.R.S. § 36-322 to be filed with the Department.

14. "Newborn screening laboratory" means a laboratory with which the Department contracts to conduct testing of the newborn screening specimens.

15. "Newborn Screening Program" means the administrative and coordination program of the Department, the newborn screening laboratory, and the follow-up services provided by designated clinical service providers.

16. "Newborn Screening Tests" means the laboratory procedures performed on a sample of blood to detect the presence of metabolic disorders and hemoglobinopathies as stated in R9-14-502.

17. "Phenylketonuria or P.K.U." means a congenital metabolic disorder characterized by abnormal phenylalanine metabolism which causes mental retardation if not treated early in life.

18. "Repeat test" means laboratory procedures performed on a specimen of blood to verify an abnormal result reported on the initial or second screen.

19. "Second screen" means laboratory procedures performed on a second specimen of blood if the initial specimen was collected within 24 hours of birth.

20. "Sickle cell diseases" mean a group of hemoglobinopathies characterized by the distortion of the red blood cells which may lead to septicemia or death in infancy if not adequately treated.

21. "Specimen collection kit" means a filter paper kit that is either licensed or approved by the Food and Drug Administration and has been approved by the newborn screening laboratory.

22. "To order" means to direct appropriate personnel to obtain specimens and send them for laboratory tests.

23. "Unsatisfactory specimen" means any blood sample rejected by the Newborn Screening Laboratory, prior to testing, that could provide unreliable, misleading, or clinically inaccurate results.

24. "Working day" means 8:00 a.m. to 5:00 p.m. Monday through Friday excluding state holidays

A. The attending physician or other person required to make a report on the birth of a newborn born in Arizona shall order or cause to be ordered the following tests for metabolic disorders and hemoglobinopathies: phenylketonuria, galactosemia, congenital hypothyroidism, biotinidase deficiency, homocystinuria, maple syrup urine disease, sickle cell disease, and other hemoglobinopathies. If a parent or guardian refuses the newborn screening tests, such refusal shall be documented in writing and shall be part of the newborn's medical record with a copy sent to the Newborn Screening Program.

B. If the initial screening sample was collected within 24 hours of birth, the responsible person shall inform the newborn's parents or guardian that a second screen for metabolic disorders shall be performed between 3 and 7 days of age.

C. The specimens for testing shall be sent, no later than 24 hours or the next working day after being obtained, to the newborn screening laboratory.

D. The results from all abnormal newborn tests for metabolic and/or hemoglobin disorders shall be reported to the Newborn Screening Program, which shall notify the health care provider.

E. The results from any confirmatory testing, ordered in response to an abnormal newborn screen, shall be reported to the Newborn Screening Program.

R9-14-503. Persons and Health Care Facilities Responsible for Tests

A. Births occurring in a health care facility.

- 1. The health care provider shall order the tests or shall ensure that the tests are ordered.**
- 2. The administrator in charge of the health care facility or the administrator's designee shall ensure that specimens are collected on all newborns born or transferred to that health care facility. The newborn's medical record shall indicate that the tests were ordered or that the parent or guardian refused the test.**
- 3. The specimens for these tests shall be obtained when the newborn is three days of age or immediately prior to the time of discharge from the health care facility, whichever is earlier.**
- 4. The specimen shall be capillary or venous blood and shall be collected utilizing a specimen collection kit obtained from the newborn screening laboratory. Cord blood shall not be accepted.**
- 5. The person in charge of the health care facility or the designated representative shall ensure that all information requested on the form within the specimen collection kit is completed.**
- 6. If the newborn is transferred to another health care facility before 3 days of age, the receiving health care facility shall be responsible for obtaining the specimen for newborn screening.**
- 7. If the initial screening specimen for any newborn was collected within 24 hours of birth, the health care facility and the health care provider shall inform the newborn's parents that a second screen is required at 3 to 7 days of age.**

B. Births occurring outside a health care facility.

- 1. The health care provider shall order the tests or shall ensure that the tests are ordered.**

2. If the initial screening sample for any newborn was collected within 24 hours of birth, the health care provider shall notify the newborn's parents that a second screen is required between 3 and 7 days of age.

3. If the birth is not attended by a health care provider with the authority to order the test, the person required by A.R.S. § 36-322(E)(3) or (4) to report the birth shall notify the local or state registrar when the certificate of live birth is filed. The registrar shall notify the health officer in the county where the newborn's parents are expected to reside. The health officer shall ensure collection of a specimen within three days from the time of notification of the birth.

4. The specimen shall be capillary or venous blood and shall be collected utilizing a specimen collection kit obtained from the newborn screening laboratory. Cord blood shall not be accepted.

R9-14-504. Parent or Guardian Education

A. The health care provider or designee shall inform the newborn's parent or guardian of the reasons for the tests.

B. The health care facility shall be responsible for distributing written educational materials on newborn screening provided by the Department.

R9-14-505. Screening Fees; Collection

The following fees shall be charged for newborn screening:

1. The fee shall be \$20.00 for an initial test.

2. The fee shall be \$15.00 for a second screen.

B. There shall be no fee charged for a repeat test or for an unsatisfactory specimen.

APPENDIX G

CHILDREN'S REHABILITATIVE SERVICES MEDICALLY ELIGIBLE CONDITIONS

ARTICLE 3. MEDICALLY ELIGIBLE CONDITIONS

R9-7-301. Medically eligible conditions; ineligible conditions

A. Applicants with the following medical conditions shall be eligible for treatment by the CRS program:

- 1. Cerebral palsy.**
- 2. Cleft lip and cleft palate.**
- 3. Myelomeningocele (spina bifida).**
- 4. Cystic fibrosis.**
- 5. Neurofibromatosis.**
- 6. Metabolic diseases:**
 - a. Phenylketonuria.**
 - b. Galactosemia.**
 - c. Homocystinuria.**
 - d. Hypothyroidism.**
 - e. Maple syrup urine disease.**
 - f. Biotinidase deficiency.**
- 7. Scoliosis.**
- 8. Sick cell anemia.**
- 9. Cardiovascular system disorder:**
 - a. Congenital heart disease.**
 - b. Cardiomyopathies.**
 - c. Valvular disorders.**
 - c. Arrhythmias**
 - d. Conduction defects.**
 - e. Rheumatic heart disease which is not in acute stage.**
 - f. Renal vascular hypertension, catecholamine hypertension.**
 - g. Arteriovenous fistulas.**
 - h. Kawasaki disease which is not in the acute stage.**
- 10. Endocrine system disorders:**
 - a. Hypothyroidism.**
 - b. Hyperthyroidism.**
 - c. Adrenogenital syndromes.**
 - d. Addison's Disease.**
 - e. Hypoparathyroidism.**
 - f. Hyperparathyroidism.**
 - g. Panhypopituitarism.**
 - h. Diabetes insipidus.**
- 11. Genito-urinary system disorders:**
 - a. Vesicoureteral reflux, chronic.**
 - b. Ectopic ureter.**
 - c. Ambiguous genitalia.**
 - d. Ureteral stricture.**
 - e. Hypospadias, complex.**
 - f. Obstructive uropathy, hydronephrosis.**
 - g. Deformity and dysfunction secondary to trauma.**
 - h. Pyelonephritis which has failed medical management and requires surgical intervention.**

12. **Ear, nose, and throat disorders:**
 - a. **Cholesteatoma.**
 - b. **Chronic mastoiditis.**
 - c. **Deformity and dysfunction secondary to trauma.**
 - d. **Neurosensory hearing loss.**
 - e. **Congenital malformations.**
 - f. **Significant conductive hearing loss equal to or greater than 30 decibels, pure bone average, which, despite medical treatment, requires hearing augmentation device.**
13. **Musculoskeletal system disorders:**
 - a. **Osteochondrodysplasias:**
 - i. **Achondroplasia.**
 - ii. **Diastrophic.**
 - iii. **Dwarfism.**
 - iv. **Larsen Syndrome.**
 - b. **Juvenile rheumatoid arthritis and seronegative spondyloarthropathies.**
 - c. **Orthopaedic complications of hemophilia.**
 - d. **Neuromuscular disorders:**
 - i. **Progressive muscular dystrophy.**
 - ii. **Arthrogryposis multiplex congenita.**
 - iii. **Spinal muscular atrophy.**
 - e. **Bone and joint infections in a chronic stage.**
 - f. **Upper limb malformations:**
 - i. **Amputations.**
 - ii. **Syndactyly.**
 - g. **Spinal deformity:**
 - i. **Idiopathic scoliosis.**
 - ii. **Congenital spine deformity.**
 - iii. **Scheuermann's Disease.**
 - iv. **Spondylolisthesis.**
 - h. **Cervical spine abnormalities, congenital and developmental.**
 - i. **Lower limb malformation:**
 - i. **Leg length discrepancies.**
 - ii. **Congenital deformity.**
 - iii. **Amputations.**
 - j. **Collagen and vascular diseases.**
14. **Gastrointestinal system disorders:**
 - a. **Tracheoesophageal fistula.**
 - b. **Anorectal atresia.**
 - c. **Hirschsprung's Disease.**
 - d. **Diaphragmatic hernia.**
 - e. **Gastroesophageal reflux which has failed medical management and requires surgical intervention.**
 - f. **Deformity and dysfunction of at least three months duration secondary to trauma.**
 - g. **Biliary atresia.**
 - h. **Congenital atresia, stenosis, fistuli or rotational abnormalities of the gastrointestinal tract.**

- i. **Omphalocele after gastroschisis.**
- 15. **Nervous system disorders:**
 - a. **Uncontrolled seizure disorders where there has been more than two seizures with documented adequate blood levels of one or more medications.**
 - b. **Seizure disorders, simple or controlled, only when the enrollee is not covered by AHCCCS or private insurance.**
 - c. **Myopathies and muscular dystrophies.**
 - d. **Myoneural disorders.**
 - e. **Neuropathies, hereditary and idiopathic.**
 - f. **Central nervous system degenerative diseases.**
 - g. **Central nervous system malformations and structural abnormalities.**
 - h. **Hydrocephalus.**
 - i. **Craniosynostosis of the sagittal or unilateral coronal sutures of a child less than 18 months of age.**
 - j. **Myasthenia gravis, congenital or acquired.**
 - k. **Benign intracranial tumor.**
 - l. **Benign intraspinal tumor.**
 - m. **Residual dysfunction after resolution of an acute stage of vascular accident, inflammatory condition or infection of the central nervous system.**
 - n. **Tourette's Syndrome.**
 - o. **Trigonocephaly, with evidence of intracranial pressure as determined by medical review by the regional medical director.**
- 16. **Ophthalmologic disorders:**
 - a. **Cataracts.**
 - b. **Glaucoma.**
 - c. **Disorders of the optic nerve.**
 - d. **Disorders of the lacrimal duct system. AHCCCS members shall be treated through an AHCCCS provider.**
 - e. **Retinopathy prematurity.**
 - f. **Disorders of the iris, ciliary bodies, retina or lens.**
- 17. **Respiratory system disorders which manifest themselves as anomalies of larynx, trachea, and bronchi and which require surgical intervention.**
- 18. **Dermatologic disorders which are medically confirmed by the CRS Regional Medical Director:**
 - a. **Craniofacial anomalies which require multidisciplinary treatments.**
 - b. **Burn scars which are functionally limiting.**
 - c. **Microtia which are grossly deforming.**
 - d. **Macrotia which are grossly deforming.**
 - e. **Complicated nevi requiring staged procedure.**
 - f. **Hemangioma of functional or diagnostic concern.**
 - g. **Craniosynostosis.**
- 19. **Genetic and metabolic disorders:**
 - a. **Amino acid and organic acidopathies.**
 - b. **Inborn errors of metabolism.**
 - c. **Storage diseases.**

20. Dental disorders:

- a. Documented significant functional malocclusion.**
- b. Enrollees with shunts.**
- c. Cardiac enrollees at risk for septic bacterial endocarditis.**
- d. Treatment-related problems (dilantin hyperplasia), for seizure disorder.**

- B. Any medical condition not specifically designated as eligible in this section or in R9-7-302 is ineligible for treatment by CRS.**

R9-7-302. Special medical conditions

- A. Both children and adult CRS enrollees shall be covered for all manifestations and complications usually associated with cystic fibrosis and sickle cell anemia.**
- B. Adults with cystic fibrosis or sickle cell anemia who are AHCCCS eligible shall not be eligible for CRS services.**

APPENDIX H

NEEDS ASSESSMENT RESEARCH QUESTIONS

I. Genetics Services For Individuals Considering Parenthood (1-30)

A. Description of Population at risk.

1. How many women of childbearing age?
2. How many pregnancies per year?
3. How many pregnancies through fertility clinics (CY 1993,1994)?
4. How many adoptions?
5. What are the preventable (through preconception intervention) birth defects?

B. Clinical Services

6. Are there family planning clinics?
7. How many women of child bearing age use Family Planning clinics?
8. How many infertility clinics?
9. How many adoption agencies?
- 9.1 Is any title XX funding used for genetic services?

C. Laboratory Services

No questions

D. Referral System and tracking

10. If families are being identified to be "at risk" (by family planning and infertility clinics): is genetic counseling provided by clinic personnel or are they being referred to genetic services?

E. Education

11. Are genetic risks being assessed in family planning clinics, infertility clinics (for donors and couples) or adoption agencies through the provision of pamphlets or questionnaires?
 - 11.1 Are there any written guidelines recommending literature or a preconception questionnaire to assess genetic risks?
 12. Is any literature being provided on smoking, drinking and drugs in the family planning clinics?
 - 12.1 Are there any written guidelines recommending the provision of these pamphlets?
 13. Has any training in genetic disorders (including pedigree taking and where to obtain consultation) been provided to personnel?
14. Has there been any organized effort to address folic acid supplementation for women who have had a previous child with NTD?

F. Consumer issues

15. What is the general knowledge of genetics in the general population?

G. Social, Legal and Ethical Implications

16. Is there any legislation regarding family planning or adoption agencies and does it address any genetic issues?

H. Databases

17. Are there any clinical database for family planning?
18. If so, does this databases include any questions regarding genetic services?

II. Genetic Services for Pregnant Women (31-100)

A. Description of population "at risk".

31. How many births are there in Arizona and what percent result in birth defects?
32. How many births result in Neural Tube Defects?

B. Clinical Services

Prenatal Genetic Diagnosis

33. Which physician groups provide prenatal genetic screening and testing services?
33.1 What procedures do they offer?
34. What are the credentials of these physicians?
35. Do these physicians work with board certified geneticists or genetic counselors?
36. What percentage of pregnant women receive the procedures (amniocentesis, CVS, PUBS etc.) for advanced maternal age, abnormal MSS, single gene, family history of a chromosomal abnormality or abnormal ultrasound?
37. What percentage of women 35 years or older are offered amnio/ CVS?
38. What percentage of women 35 years or older receive the procedure?
39. What is the economic, demographic and insurance breakdown of women who receive procedures? (i.e. How many are Hispanic or Native American?)
40. Do HMOS, Medicaid, insurance companies pay for these services and or reimburse at a low or limited rate?
41. Does IHS and other federal programs provide or pay for these services?
41.1 Are there any groups of women that have no access to services for any reason?

Level II Ultrasonography

- 42. How many physician groups provide level II ultrasonography?**
- 43. What are the credentials of these physicians?**
- 44. How many perform fetal echocardiography?**
- 45. How many result in an invasive procedure?**
- 46. Who pays for these procedures?**

Maternal Serum Screening

- 47. What percentage of pregnant women have MSS?**
- 48. What percentage are offered MSS?**
- 49. Do most receive (or are offered) MSAFP or multiple marker?**
- 50. What is the socioeconomic and demographic breakdown of women who receive MSS?**
- 51. What percentage of insurance companies reimburse for MSS?**

Teratogen

- 52. How many pregnancies are referred to geneticists for teratogen counseling?**
- 53. Is there a statewide teratogen counseling hotline?**

C. Laboratory Services

Cytogenetics

- 54. Which laboratories are specimens sent to?**
- 54.1 What types of Cytogenetic testing are done?**
- 55. Do the laboratories meet all state and or national guidelines?**
- 56. How many specimens are sent out of state?**
- 57. Who pays for these services?**
- 58. Does IHS, Medicaid or other federally funded programs pay for these services?**

Molecular and Biochemical

- 59. Where are the specimens being sent for DNA analysis?**
- 60. Where are specimens being sent for biochemical (AFP, acetylcholinesterase etc) and other testing?**

Maternal Serum Screening

- 61. Which laboratories are the specimens sent to?**
- 62. Are any of the specimens sent out of state?**
- 63. Are all in-state laboratories performing the triple marker test?**

D. Referral System and Tracking

- 64. Are physicians and laboratories monitoring pregnancy outcomes?**
- 65. Of those who deliver children with NTD, how many have had MSS?**

E. Education

- 66. Has any training in genetics been provided to OB-Gyn physicians and other health providers that work with pregnant women?**
- 67. What is the level of knowledge among OB providers on MSS ?**
- 68. What is the level of knowledge among consumers on MSS?**
- 68.1 Has there been any coordinated educational effort to prevent Fetal Alcohol Syndrome.**

F. Consumer issues

- 69. What issues do women have regarding MSS?**
- 70. What issues do women have regarding prenatal diagnosis?**

G. Social, legal and ethical questions

- 71. Is there a perception among the genetic and laboratory providers to standardize MSAFP programs?**
- 72. Is there a perception to mandate MSS screening?**
- 73. Do prenatal providers offer CVS/amnio for the purpose of sex selection?**
- 73.1 How many pregnancies are terminated due to a detection of a fetal abnormality?**
- 73.2 Do most Health Plan contracts (Insurance, state and federal funding) reimburse for termination of pregnancy?**

H. Databases

- 74. Do you participate with MSRGSN clinical service database. If so, for how long and is data complete for CY 1993 and 1994.**
- 74.2 Does the provider maintain any other type of database?**

III. Genetic services for children and their families (101-160)

A. Description of population "at risk"

- 101. What is the birth defects rate in Arizona? estimate for 1993 and 1994)**
- 102. What is the birth defects rate per county? (estimate for 1993, and 1994)**
- 103. What percentage of these children with a birth defect be consider to be at risk for a genetic disorder?**
- 104. What is the prevalence of specific single gene disorders that are not identified through the birth defects registry?**

B. Clinical Services

Genetic Clinics

- 105. What percentage of infants identified through the Arizona Birth Defects Monitoring Program (ABDMP) are receiving genetic services?**
- 106. Who provides genetic services for the pediatric population?**
- 107. Where are genetic services being provided?**
- 108. Do insurance and HMO pay for genetic services?**
- 109. What are CRS financial and medical eligibility criteria?**
- 110. What percentage of children identified at risk for genetic disorders are eligible for genetic services through CRS? (Maybe identify top 5 conditions that are not eligible for CRS)**
- 111. Are we serving all children with genetic disorders?**
- 112. Who is referring to genetic services?**
- 113. What is the economic, demographic and insurance breakdown of families that are served? (How many Hispanic and Native Americans are being served?)**
- 113.1 Is there a need for the continuation of genetic clinics in reservation sites?**
- 113.2 Is there a need to establish clinics for the Hispanic population?**
- 113.3 What is the current status of CRS field clinics (genetics)?**
- 114. What are the most common reasons for referrals?**

Specialty clinics

- 115. Who provides treatment for individuals with specific genetic conditions (e.g. metabolic, hemophilia etc)?**
- 116. How many and what type of specialty clinics are there?**
- 116.1 Is there a need to establish screening clinics for Fetal Alcohol Syndrome within the reservation System?**
- 117. Do HMO, insurance companies, Medicaid or other federal (e.g. IHS) or state supported programs pay for these services?**
- 118. What are CRS medical and financial eligibility criteria for their specialty clinics?**
- 119. Do these clinics serve both children and adults?**

Family Genetic Testing

- 120. Is carrier testing through DNA analysis available for families for Cystic Fibrosis, hemophilia, and Duchenne Muscular dystrophy?**
- 121. Do insurance companies (or other health plans) pay for these services?**
- 122. Is there carrier testing for other disorders available to families?**
- 123. If so, Who pays for these services?**

C. Laboratory Services

- 124. What laboratories within the state are providing genetic testing karyotype, screening for metabolic disorders, enzyme analysis and DNA analysis)?**

- 125. How many specimens are sent out of state?
- 126. How many laboratory tests are ordered by non-geneticists?

D. Referral System and Tracking

- 127. Are families coming back to genetic services for follow-up?
- 128. Is duplication of services occurring?(Same patient seen in Maricopa and CRS?)
- 129. Do families have access to other services such as early intervention programs or developmental disabilities programs? (Do genetic providers refer them to these programs?)
- 130. Do children have access to primary care services?

E. Education

- 131. How is the public being informed of genetic services?
- 132. How are the medical community and other health professionals being informed on genetic services?
- 133. How much genetic education is being provided to physicians in specialties such as pediatrics, family practice and internal medicine?

F. Consumer issues (Parent Forum)

- 134. What are the barriers to obtaining existing services?
- 135. What are the gaps of current services?
- 136. What are the problems with existing services?
- 137. What currently works?

G. Social Legal and Ethical implications

- 138. Are there laws within the state that mandate that families of children with birth defects can obtain insurance?
- 139. Is gene therapy for some genetic disorders provided within the state of Arizona?
- 139.1 Is there legislation addressing patient confidentiality?

H. Databases

- 140. Are there any clinical databases for clinical genetic services?
- 141. Are there any clinical genetic services for specialty clinics?

IV. Genetic Services for Adults with genetic disorders (161-200)

A. Description of Population "at risk"

- 161. What is the prevalence of adults with genetic disorders? (early and late onset).
- 162. What is the prevalence of each of the following genetic conditions: genetic colon cancer, genetic breast cancer and Huntington disease?

B. Clinical services

Genetic Clinics

- 163. Are adults with genetic disorders currently being served in the genetic clinics for either diagnosis or treatment?
- 164. Do insurance companies and HMOs reimburse for these services?
- 164.1 To whom or where are adults being referred to from the pediatric genetic clinics?

Specialty Clinics

- 165. Are all the specialty clinics serving adults?
- 166. Is there a need for other specialty clinics to serve children and or adults?
- 166.1 Are all specialty clinics who serve adults serving an appropriate number of adults?
- 166.2 Are internal medicine and family practice physicians serving adults with genetic disorders in there general clinics?

Pre-symptomatic services

- 167. What kinds of services for pre-symptomatic testing of adults are being provided by genetic providers?
- 168. For what disorders? (breast cancer, colon cancer, Huntington, etc.)
- 169. Which health providers are currently providing these services (geneticist or other)?

C. Laboratory Services

- 170. Are geneticists sending specimens for presymptomatic testing of colon cancer, breast cancer and Huntington disease?
- 170.1 What type of physician (geneticists?) are sending specimens for presymptomatic testing?
- 171. Are insurance companies paying for these services or are they considered experimental?

D. Referral system and tracking

No questions

E. Education

No questions

F. Consumer Issues

172. What are the consumer issues? (Family forum)

172.1 Should a forum be held for adults with genetic disorders?

G. Social, Legal and Ethical implications

173. Once an individual is identified presymptomatically, what are the issues regarding confidentiality and insurability?

174. Who should be providing presymptomatic testing to families?

175. Who should be establishing clinical practice standards for presymptomatic testing as new technologies allow for the identification of more genetic disorders?

H. Databases

No questions

V. Population Based (Public Health) (201-270)*

NOTE: Since one of the goals of the state genetics plan is to determine what the role of Public Health is in the administration, planning, and implementation of genetic services; it was decided to integrate the questions under this category (population based) into the other four categories. This will decrease any bias that can occur based on the services that are currently being provided through the ADHS. However, for "working purposes" this category will remain until after the implementation of the needs assessment.

A. Description of population "at risk"

Newborn Screening (Newborns)

201. What is the incidence and prevalence of all seven disorders?

201.1 What are the disorders?

Hemoglobinopathy Screening (other than newborns)

202. What is the incidence or prevalence of hemoglobinopathies?

Genetic service programs

203. Incidence and prevalence of birth defects (Repeat from IIIA)

Registries

204. Incidence and Prevalence of Birth defects (Repeat from IIIA)

205. Description of Cancer Registry.

205.1 Description of child Fatality review. Specifically regarding infant mortality due to congenital anomalies and neoplasms.

B. Clinical Services

Newborn Screening

- 206. Does the state or other or other funding sources pay for metabolic formula, penicillin or Synthroid?**
- 207. Do HMOs or insurance companies pay for these clinical services?**
- 208. Do HMOs or insurance companies pay for metabolic formula, Synthroid or penicillin?**

Hemoglobinopathy Screening (carrier testing)

- 209. Who is screening the "Non-newborn" population (carrier detection for sickle cell disease) and where are the specimens being sent to?**
- 209.1 Is carrier detection for traits other than C or S being offered to the "non-newborn" population?**
- 210. Who provides genetic counseling?**
- 211. Who pays for both screening and counseling?**

Genetic service programs

- 212. Form what sources are funds available for CRS?**
- 213. What is the medical and financial eligibility criteria to obtain genetic services through CRS?**
- 214. Do MCH block or other federal/state monies fund genetic services?**
- 214.1 What is the role of MCH in the planning administration and funding of genetic services?**
- 214.2 Are there any other genetic programs administered through federal an/or state funding?**

C. Laboratory Services

Newborn Screening

- 215. Is there a centralized lab that handles all the newborn screening specimens?**
- 216. Does this centralized lab meet national standards ?**
- 217. Does the lab participate in a quality assurance program?**
- 218. Who is paying for confirmatory testing?**
- 219. Is confirmatory testing done in a timely manner?**
- 220. Is confirmatory testing funded by the state?**
- 221. Should the state screen for other disorders? (CAH or CF)**
- 222. Should the state continue to test for all the seven disorders?**

Hemoglobinopathy Screening

- 223. Which lab provides carrier screening?**
- 224. Does this lab identify trait status other than for hemoglobin S?**
- 225. How are these lab services paid for?**

Genetic Service Programs

- 226. What laboratory services does CRS fund for patients seen in the genetic clinics?**
- 227. What other laboratory service issues are there?**

D. Referral System and Tracking

Newborn Screening

- 228. Are all newborns in Arizona screened for all seven disorders?**
- 228.1 Are all newborns who were screen before 48 hours, receiving a second screen? If so which labs are they being sent to?**
- 228.2 Are children who move into Arizona from other states receiving services?**
- 228.3 How long (days) between initial screen and beginning of treatment?**
- 229. Are all newborns with metabolic disorders (other than congenital hypothyroidism) identified through the newborn screening program receiving services through the CRS metabolic clinics?**
- 230. Are all newborns with congenital hypothyroidism receiving service through the CRS endocrinology clinics?**
- 231. Are all newborns with a hemoglobinopathy receiving services through the CRS Sickle Cell Clinics?**
- 232. Does the state plan to establish a newborn screening registry of confirmed cases to allow for tracking of these children?**
- 233. Is a centralized follow-up program being established to monitor the NBS program including the reporting of positive screens and tracking of identified cases?**

Hemoglobinopathies

- 234. Once carriers are identified are all the families being offered genetic counseling and more testing for other family members?**

E. Education

Newborn Screening

- 235. Is there an organized effort to provide education to all submitter on the newly established NBS program?**

Hemoglobinopathies

- 236. Is there any organized effort to provide education to the public regarding Sickle Cell Screening?**

Genetic Service Programs

- 237. What public education is being provided to educate the community on genetic services?**

G. Social, Legal, Ethical

- 238. What are the current Arizona laws for all the public health programs and do they apply to genetics?**
- 238.1 How is informed consent being addressed for the newborn screening program?**
- 238.2 Is there any policy within ADHS regarding the storage of filter paper specimens and the use of these specimens for future research?**
- 238.3 Are families consenting for hemoglobinopathy carrier detection when consenting for newborn screening?**

H. Databases

- 239. What are the current databases used for all the public health programs which relate to birth defects and genetic conditions?**

I. Investigational

Registries

- 240. Should the ABDMP be used to advance knowledge in the etiology or causation of birth defects.**
- 241. What resources are available or being utilized to address etiology?**
- 242. How do we use the ABDMP for the planning of services or the identification of families at risk for secondary disability?**
- 243. Should we even care that the environment may be causing some birth defects?**
- 244. How should the governmental agencies (including ADHS) address these issues.**
- 245. How should the information be used?**

Note:

Appendix H and I include planning information that was utilized by Arizona to develop the Genetic Health Services Plan. Additional information regarding the development of the Genetic Health Services Plan is available upon request from the Arizona Department of Health Services, Office of Women's and Children's Health, Genetics Program Consultant, 2927 N. 35th Ave., Phoenix, Arizona 85017.

APPENDIX I

GENETICS PLAN SUMMARY

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
2.0	Persons to have appropriate genetic risk assessment and Intervention for the prevention of birth defects and genetic Disorders.			
2.1	Obtain and review risk assessment tools.	#2 Priority	Prenatal	Jun-98
2.2	Develop a plan to promote inclusion of a statewide genetic screening component into aspects of well-woman care and preventive health services. Identify programs and agencies for collaborative links.		Prenatal	Oct-99
2.3	Develop and strengthen collaboration and links with programs and agencies		Prenatal	Ongoing
2.4	Develop a plan with other agencies and cytogenetic testing providers to assess actual utilization of prenatal chromosomal diagnosis in 35 and older.	Low priority	Prenatal	Dec-02
2.5	Monitor the Arizona network for genetic counseling services.	List in booklet “Genetic Services in Arizona”	Prenatal	Booklet printed 3/00

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
3.0 All individuals with a family history of a genetic disorder to be referred for genetic counseling and evaluation				
3.1	Promote insurance company awareness of the benefits of promoting preventive genetic services	#1 Priority: Obtain set of slides by MSRGSN Finance committee (in progress)	Clinical (Prenatal)	Ongoing
3.2	Develop a training plan for Arizona providers in genetic services and referrals		Clinical (Prenatal)	Ongoing
3.3	Convene a committee or task force to develop additional information and data about the incidence of persons in Arizona who are referred for genetic counseling and evaluation due to a family history of a single gene disorder.		Clinical (Prenatal)	Ongoing, 12-01-98
3.4	All cases with a confirmed disorder identified by Newborn Screening Program to be referred for care with a specialist		N B S	Ongoing
3.5	Support efforts to develop standard of care for disorders identified by the NBS Program		N B S	Ongoing

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
4.0 Genetic Specialists that are available, accessible, and geographically appropriate to the at-risk population				
4.1	Involve community and tribal leaders, in designing education and information programs that are community-based, and accessible to the rural and tribal communities		Clinical Committee	Oct-98
4.2	Create a task force to identify options to enhance the availability and accessibility of genetic services		Clinical Committee	As needed
4.3	Continue to strengthen links between geneticists, providers and incorporate other related agencies and teams to the network		Clinical Committee	Ongoing
4.4	Continue promotion of genetics consultation contracts through ADHS		Clinical Committee	Ongoing
4.5	Develop information for Arizona health care providers and consumers on the availability of genetic clinics, names, and addresses of all genetic service providers in Arizona.	#2 Priority: Edit and update booklet "Genetic Services in Arizona"	Clinical/ Educ	Booklet printed 3/00
4.6	Develop a plan to refine and clarify availability and accessibility standards for the genetics population		Clinical Committee	Aug-99

4/13/00

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Task	Lead	Target Completion
5.0 Genetic diagnostic procedures and genetic tests to be performed following comprehensive genetic evaluation by appropriately trained specialists				
5.1	Prepare and disseminate information outlining what consumers should look for in seeking genetic counselors and genetic testing services	MSRGSN website & brochure Newspaper/newsletter articles Develop ADHS website on genetics—link w/MSRGSN site	Education	Info for website to DHS webmaster 3/00
5.2	Prepare and disseminate lists of resources available to all providers regarding genetic testing	Use Ethical, Legal, and Social Issues of Human Genome Proj. as a resource; Update “Genetic Services in Arizona”	Education	In process; booklet printed 3/00

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
6.0 Health care providers to be knowledgeable and informed about genetic risks, services, and resources.				
6.10	Goal 1 – Improve lab submission for newborn screening/referral for other genetic tests.		Lab-general, Lab-NBS	
6.11	Collect data regarding number of unacceptable specimens currently submitted.		Lab-general	Mar-98
6.12	Prepare a genetic test submission manual containing laboratory testing requirements for common genetic tests and newborn screens		Lab-general	Mar-98
6.13	Develop and implement a training plan for educating health care providers regarding specimen submittal. Train providers in the proper submission of specimens.		Lab-general	June-98
6.14	Develop or enhance ongoing reporting or data collection mechanism for tracking unacceptable specimens		Lab-general	June-98
6.15	Investigate compliance rate of lab submission by providers for NBS and develop plan for increasing rate.		Lab-NBS	
6.16	Prepare an NBS practitioner's manual containing laboratory testing requirements for NBS		Lab-NBS	Periodic
6.17	Develop and implement a training plan for educating health care providers regarding specimen submittal for NBS	Train providers in the proper submission of specimens	Lab-NBS	Ongoing

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
6.18	Improve quality of submission by providers	(1) Collect date, provide feedback to submitters, monitor changes regarding number of unacceptable specimens submitted; (2) Collect data, provide feedback to submitters, monitor changes in incomplete data submission. (Lab QA plan for NBS);(3) Develop or enhance ongoing reporting or data collection mechanism for tracking unacceptable specimens; (4) Educate.	Lab-NBS	
6.20	Goal 2—Promote early identification and detection of need for genetic testing and services through provider education of genetic risks, community awareness.	Lectures at medical & nursing schools Update “Genetic Services in Arizona”	Education Committee	Ongoing Booklet printed 3/00
6.21	Develop and strengthen collaboration and links with programs/agencies	Update “Genetic Services in Arizona” Including but not Limited to: HIS, MSRGSN, CRS, WIC, AHCCCS	Education N B S	In process; booklet printed 3/00
6.211	Collect information about how referral processes to genetic services are initialed		Education Committee	June-98
6.212	Identify continuing education and inservice opportunities for physician providers in Arizona.	List of presentations to state genetics coord. – eval gaps	Education Committee	June-98
6.213	Develop professional provider education program for preconception, prenatal, children, and adults.		Education Committee	
6.214	Educate medical directors of health plans on the need for primary care providers to refer patients for genetic diagnostic services.	Met w/AHCCCS directors Coordinate meeting w/AHCCCS, MCH directors	Education Committee	
6.215	Educate providers about appropriate referrals and genetics health care providers.	Update “Genetic Services in Arizona”	Education Committee	Booklet printed 3/00

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
6.216	Promote inclusion of information about the NBS program in health/medical education curricula.		Lab-NBS	Jul-00
6.217	Improve provider awareness of resources available related to the disorders identified by NBS.		Lab-NBS	Ongoing
6.218	Improve provider referrals for genetic services for disorders identified by NBS.		Lab-NBS	Ongoing
6.22	Path B: Collaborate with consumer groups and parent/child advocacy organizations to educate providers about the availability of family support organizations.		Education Committee	
6.221	Survey health service, social service, nutrition, and medical ed/training programs in Arizona to obtain information about genetics curriculum content.	Obtain syllabi of genetics portion of each training program to evaluate their curricula	Education Committee	
6.222	Promote inclusion of enhanced genetics component in health/medical education curricula.		Education Committee	Ongoing
6.223	Link with others to identify opportunities for collaboration.		Education Committee	Ongoing
6.224	Based on information and assessment, develop a plan (with educational institutions to target and improve areas in need of additional coverage in the curriculum).		Education Committee	Mar-99

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
7.0 Genetic Laboratory Services to be of High Quality				
7.10	Improve availability of laboratory services		Lab-general Lab-NBS	
7.11	Explore options for development and support of additional laboratory diagnostic testing capability.		Lab-general Lab-NBS	
7.12	Identify and document existing networks of genetic laboratory providers and facilities used by AZ health care providers and organizations. Make this list available to the provider community through the education and orientation process.	Updating "Genetic Services in Arizona"	Lab-general	Booklet printed 3/00
7.13	Evaluate the current system, including specimen transportation, testing time, cost, protocol, interpretation of results, and other issues as identified.		Lab-general Lab-NBS	Dec-98
7.14	Improve availability of all types of NBS tests in Arizona	Identify: what sites are available? What is available? Steps to add services which are not available.	Lab-NBS	Ongoing
7.15	Report to Genetic Services Advisory Committee about NBS activities		Lab-NBS	Periodic
7.20	Assure mechanism for access to genetic lab testing when indicated, to ensure payment for genetic laboratory testing is available to all who require it.		Lab-general	

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
7.21	Convene a working group of the GSAC to explore options for payment, include clinicians, finance, MCO representation, AHCCCS .		Lab-general	Dec-98
7.30	Promote appropriate accreditation for laboratory services who provide genetic screening and testing services for AZ clients.		Lab-general	
7.31	Develop network and links with laboratory regulation agencies for genetic lab testing.		Lab-general Lab-NBS	Dec-98
7.32	Assure appropriate accreditation for NBS laboratory services	Develop network and links with laboratory regulation agencies for performance testing.	Lab-NBS	Ongoing
7.33	Research current requirements for laboratory certification in Arizona, determine availability of other special requirements for laboratories that handle metabolic or genetic testing specimens.		Lab-general	Dec-98
7.34	Promote use of standards of practice used by cytogeneticists and consultants to laboratories that provide genetic testing services through inclusion in existing state contracting systems (e.g., AHCCCS).		Lab-general	Dec-98
7.35	Promote use of standards of practice used by cytogeneticists and consultants to laboratories that provide genetic testing services through inclusion in existing state contracting systems (e.g., AHCCCS).		Lab-general	Dec-98

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
8.0 Increase awareness of genetic issues and genetic services				
8.10	Goal 1 – Collaborate with providers, insurance companies, and consumers to document a plan for informing consumers on how to access genetic services.		Education Lab-NBS	Ongoing
8.11	Gather information from insurance companies regarding the availability of genetics education programs.		Education Committee	Dec-98
8.12	Gather information from insurance companies, providers about how referrals to genetic services are currently initiated for preconception, prenatal, child, and adult genetic services.		Education Committee	Dec-98
8.13	Conduct focus group for input regarding needs for genetic services.		Education Committee	Mar-99
8.14	Review currently available consumer information.		Education Committee	Mar-99
8.15	Develop a statewide genetic service awareness program with input from consumers and providers.		Education Committee	Dec-99
8.16	Develop public service announcements to increase awareness of preventable genetic conditions, availability of genetic services.		Education Committee	Jun-00
8.17	Seek support for the project through March of Dimes, Federal Grant, or other funding sources.		Education Committee	Ongoing

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
8.18	Develop a statewide genetic service –NBS awareness program with input from consumers and providers.	(1) Develop and promote user friendly, easy to read written information on NBS for provider’s offices, public service announcements, and brochures; (2) Promote collaboration with other resources and teams involved in health education and preventive care; (3) Involve consumer advocacy groups, health plan member service reps, community and tribal leaders, other family advocacy agencies in designing education & information programs that are community-based and accessible to the rural and tribal communities.	Lab-NBS	

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
9.0	All individuals are informed of the benefits, risks, and limitations for genetic testing		Education Committee	
9.10	Complete accurate and clear consent information and other information for genetic testing		Education Committee	
9.11	Conduct consumer, ADHS, provider focus group		Education Committee	Mar-99
9.12	Make recommendations and suggestions for informed consent for genetic testing		Education Committee	06/31/99
9.20	Provide these materials to providers and consumers.		Education Committee	Dec-98
9.21	Gather materials currently available, disperse to focus group by 1/99		Education Committee	Mar-99
9.22	Review currently available information during focus group meeting		Education Committee	Sep-99
9.23	Develop educational materials to facilitate the provision of this instruction		Education Committee	Jan-00
9.24	Develop and implement multi-media training sessions or workshops to include opportunities for interactive practice in counseling about genetic services and testing		Education Committee	Jan-00
9.25	Provide educational materials regarding genetic services and treatment options		Education Committee	Jan-00

4/13/00

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
9.30	In a collaborative effort, develop appropriate information, and “minimum standards” regarding benefits, risks and options for genetic testing, to be shared with clients, parents, and providers.		Education Committee	
9.31	Review ACOG, American Academy of Pediatrics, Alliance of Genetic Support Groups, and other guidelines for informed consent, patient rights and responsibilities.		Education Committee	
9.32	Develop or refer to guidelines for altered medical management of genetic disorders and pregnancy in cases with altered maternal or fetal genetic conditions (e.g., caesarian section when baby has spina bifida).		Education Committee	
9.33	Review existing statements and develop a statement of client rights and responsibilities related to genetic testing services.		Education Committee	Dec-98
9.34	Provide these materials to providers and consumers.		Education Committee	Jan-99

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
10.0 All persons to be protected from discrimination resulting from genetic testing, services, or disorders.				
10.10	Educate the legislative community about genetic services. Prepare educational information in response to requests from various governmental agencies of legislative bodies.		Education Committee	Ongoing
10.11	Education Subcommittee assists to develop appropriate educational materials for legislature for newborn screening, genetic lab testing.		Education Committee	Dec-98
10.12	Recommend improvements in legislation when needed.		Education Committee	Ongoing
10.20	Promote integration of newly developed or approved legislation regarding genetic discrimination into the community.		Education Committee	Ongoing
10.21	Monitor the implementation of new legislation to the community.		Education Committee	Ongoing
10.20	Develop systems and processes to ensure that genetic information is not used to make health care and other insurance prohibitively expensive.		Education Committee	
10.31	Search for data, info on cases (through DOI and other sources), compile case data, promote/support education on this issue.		Education Committee	Dec-99
10.32	Support efforts to ensure that genetic information obtained as a result of NBS is not used to make health care and insurance prohibitively expensive.	(1) Develop appropriate educational materials for legislators for NBS; (2) Recommend improvements in legislation when appropriate.	Lab-NBS	Ongoing

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Task	Lead	Target Completion
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11.0 Improved data collection and analysis re: birth defects, diseases, Genetic services and health outcome.				
11.1	Utilize a working group of GSAC to conduct an in-depth analysis of genetic services data reporting requirements		Data Com (Birth Defects Registry)	
11.2	Develop a core set of questions and issues regarding genetic services data, such as information regarding genetic risk assessment, genetic education and utilization reporting.		Data Com (Birth Defects Registry)	
11.3	Study genetics services data and reporting requirements for all systems—AHCCCS, DES adoption, private adoption agencies, Newborn screening program CRS specialty providers, and commercial insurers.		Data Com (Birth Defects Registry)	
11.4	Determine parameters for access to genetic services data and information.		Data Com (Birth Defects Registry)	
11.5	Create a minimum data set of genetic services information, including a data dictionary (outlines definitions for data elements).		Data Com (Birth Defects Registry)	
11.6	Improved NBS data collection and analysis re: birth defects, diseases, genetic services, and health outcome.	NBS data—timing, confirmed cases, abnormal screens, % normal at first and abnormal at second screen.	Ongoing	

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
12.10	Persons to have access to single site multi-specialty, interdisciplinary teams when appropriate for care and treatment of identified genetic diseases			
12.10	Adult			
12.11	Obtain more information about adult multi-specialty, interdisciplinary clinics.		OCSHCN/CRS (Clin)	In process
12.12	Develop a task force to examine multi-specialty, interdisciplinary care options for genetic disorders. Define the task force role, develop other venues or options for multi-specialty, interdisciplinary care for adults.		OCSHCN/CRS (Clin)	In process
12.13	Identify barriers for adults to access multi-specialty, interdisciplinary care and services for genetic disorders (e.g., geography, personnel, \$, space, and transportation).		OCSHCN/CRS (Clin)	In process
12.14	Consider network expansion options such as case management models, etc. Other payer and network relationships need to be identified, including use of children's networks for care of adults.		OCSHCN/CRS (Clin)	Dec-98
12.20	Pediatrics		OCSHCN/CRS (Clin)	
12.21	Obtain more information about pediatric multi-specialty, interdisciplinary clinics.		OCSHCN/CRS (Clin)	In process
12.22	Develop a task force to examine multi-specialty, interdisciplinary care options for genetic disorders. Define the task force role, develop other venues or options for multi-specialty, interdisciplinary care for adults.		OCSHCN/CRS (Clin)	In process
12.23	Identify barriers for children to access multi-specialty, interdisciplinary care and services for genetic disorders (e.g., geography, personnel, \$, space, and transportation)		OCSHCN/CRS (Clin)	In process

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
12.24	Consider network expansion options such as case management models, etc. Other payer and network relationships need to be identified, including use of children's networks for care of adults.		OCSHCN/CRS (Clin)	Dec-98
12.30	Implementation		OCSHCN/CRS (Clin)	
12.31	Explore options with CRS for expanding medical eligibility and funding for CRS services, by diagnosis first, then expand for age after diagnosis.		OCSHCN/CRS (Clin)	
12.32	Develop a plan to improve multi-specialty, interdisciplinary services and clinics for preconception, prenatal, children and adult services.		OCSHCN/CRS (Clin)	Dec-98
12.34	Implement the plan to improve multi-specialty, interdisciplinary services and clinics for preconception, prenatal, children and adult services.		OCSHCN/CRS (Clin)	Dec-01

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
13.0 All children to have access to genetic information about their Biologic family				
13.1	Review current legal and legislative requirements for genetic information gathering for adoptions (public and private).		Clinical (Prenatal)	Completed
13.2	Review national standards or guidelines from other states regarding collection of genetic information prior to adoption.		Clinical (Prenatal)	Completed
13.3	Identify possible resources (and those already available) that could be used to assist birth parents to provide complete information (e.g., questionnaire, family history taken).	Low priority	Clinical (Prenatal)	Dec-01
13.4	Develop and implement an educational program with Adoption Agencies regarding the importance of genetic information.	Low priority	Clinical (Prenatal)	Dec-01
13.5	Work with DES, and private adoption agencies to implement these resources for improving non-identifying data collection of genetic information.	Low priority	Clinical (Prenatal)	Dec-02
13.6	Evaluate if birth parents are supplying genetic information and if it is available to the adoptive child.	Low priority	Clinical (Prenatal)	Dec-01

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
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14.0	Complete genetic background information to be collected about egg and sperm donors.
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14.1	Review current legal and legislative requirements for genetic information gathering for egg and sperm donation.		Prenatal (Clinical)	Tabled
14.2	Review national standards or guidelines from other states regarding collection of genetic information prior to egg or sperm donation.		Prenatal (Clinical)	Tabled
14.3	Identify possible resources (and those already available) that could be used to assist donors in providing complete information, and the recipients to receive it. (e.g., questionnaire, family history taken).		Prenatal (Clinical)	Tabled
14.4	Work with DES, and infertility clinics, or assisted reproduction facilities to investigate options for improving non-identifying data collection of genetic information of egg and sperm donors.		Prenatal (Clinical)	Tabled
14.5	Develop and implement an educational program, including a conference for infertility/OB providers and sperm bank staff, and educate providers to develop and implement a donor/carrier awareness program.		Prenatal (Clinical)	Tabled
14.6	Implement donor carrier testing for common genetic disorders when available and provide appropriate follow-up.		Prenatal (Clinical)	Tabled
14.7	Evaluate if donors provide information, receive carrier testing and follow- up information.		Prenatal (Clinical)	Tabled

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
15.0 All persons to exercise free choice with respect to genetic testing or Treatment.				
Core Value				
15.1	Ensure that use of genetic testing services is voluntary.		Clinical Committee	
15.2	Promote use of non-directive counseling.		Clinical Committee	Ongoing
15.3	Develop informed consent checklist (Using input and review from geneticists and consumers) for counselors and providers to use in discussing genetics testing, treatment, and options with clients and families.		Clinical Committee	Dec-02
15.4	Include translation education themes in curricula, health services education, medical CE, consumer/prenatal ed, provider training, pt. Bill of Rights, information about procedures to Lay Health Workers.		Education Committee	
15.5	Gather policy statements related to presymptomatic genetic testing in children.	#3 Priority Committee will be gathering information.	Clinical Committee	Ongoing
15.6	Improve education of providers and parents enabling parents to exercise free choice with respect.	Providing hospitals with a parent refusal form and education on statute, rules and parent right to refuse.	NBS Committee	

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Tasks	Lead	Target Completion
16.0 Genetic services to be family centered, culturally competent, Integrated, geographically appropriate, and focused on the Maintenance of health.				
Core Value				
16.01	Establish a multidisciplinary task force/subcommittee to identify strategies for improving coordination of care and services for all persons who need or receive genetic services.		Clinical Committee	Ongoing
16.02	Investigate current case management networks existing in AZ for the care and treatment of persons affected with genetic services.		Clinical Committee	Ongoing
16.03	Enhance communication and referral networks between rural primary care providers and urban based genetics, multidisciplinary treatment specialists and insurance payers.		Clinical Committee	Ongoing
16.04	Explore the possibility of increasing access to information for the public and consumers, through technology such as Medline, Internet or the ADHS Websites.	List of web sites as handout at Genetics Conf. May 12-14, 1999	Education Committee	
16.05	Continue to strengthen links with other DHS teams, DHS, and DES home visiting programs, parent support groups.		Clinical NBS	Ongoing
16.06	Work with providers and parent support groups to develop and implement a confidential satisfaction survey for persons who receive genetic screening or testing services.	Phx Perinatal Assoc. has follow up program & pt. Satisfaction survey; CRS has pt. Satisfaction survey.	Education Committee	Ongoing
16.07	Use of results of satisfaction survey to develop a plan for improvement.		OCSHCN/ CRS	Ongoing
16.08	Work with wellness and prevention programs to identify strategies for wellness promotion in persons with genetic conditions.		Clinical Committee	Ongoing

**Genetic Health Services Plan
Action Plan
SORT BY STANDARD**

Standard	Action	Ta sks	Lead	Target Completion
16.09	Identify and link with current follow-up and coordination programs, and encourage new programs where there are gaps in service.		Clinical Committee	Ongoing
16.10	Explore linking with Office of Minority Health.		Education Committee	
16.11	Prepare a Spanish translation for all written information provided to the public regarding genetic services.	MSRGSSN & CRS brochures; SPRANS Hispanic grant—ended 1998	Education NBS	Ongoing
16.12	Offer training for professionals, including medical providers, hospital personnel, and teachers in cultural awareness and sensitivity issues surrounding genetic services.	SPRANS Hispanic grant—ended 1998	Education Committee	
16.13	Promote availability of verbal translation services and service coordination to meet the needs of individuals seeking genetic/NBS services. Consider availability of translation services for tribal populations and appropriateness of information dissemination in written or spoken languages.	Translation available at CRS and many other clinics; SPRANS Hispanic grant—ended 1998	Education NBS	
16.14	Involve community and tribal leaders in designing education and information programs that are community based and accessible to rural and tribal communities.		Education Committee	
16.15	Involve consumer advocacy groups, health plan/insurance member service representatives, and other family advocacy agencies in efforts.		Education Committee	
16.16	Involve parent support groups and consumer advocates in designing materials for genetics program and the public.		Education Committee	Ongoing
16.17	Develop materials for consumers and the public that is user-friendly, and easy to read.	MSRGSSN & CRS brochures	Education Committee	Ongoing
16.18	Consider availability of translation services for tribal populations and appropriateness of NBS information dissemination in written or spoken language.		NBS	
16.19	Promote education in all sectors of the population regarding genetic contribution to common diseases as well as hereditary illnesses.		Education Committee	

**Arizona Genetics Health Services Plan
Needs Assessment Grid**

	A. Population “at risk”	B. Clinical Services	C. Laboratory Services	D. Referral & Tracking	E. Education	F. Consumer Issues	G. Social, Legal & Ethical	H. Databases	I. Investigational
I. Preconception									
Family Planning	1-5	6-7	NQ	10	11-14	15	16	17-18	NQ
Infertility	1-5	8	NQ	10	11,13,14	15	16	NQ	NQ
Adoption	1-5	9	NQ	10	11,13	15	16	NQ	NQ
II. Pregnancy									
PDX	31-32	33-41	54-60	64	66	70	73	74	NQ
Ultrasound	31-32	42-46	NQ	64	66	NQ	NQ	NQ	NQ
MSS	31-32	47-51	61-63	64,65	69	69	71,72	NQ	NQ
Teratogen	31-32	52-53	NQ	64	66	NQ	NQ	NQ	NQ
III. Children									
Genetic	101-104	105-114	124-126	127-130	131-133	134-137	138-139	140	NQ
Specialty	101-104	115-119	124-126	127-130	131-133	134-137	138-139	140	NQ
Genetic Testing	101-104	120-123	124-126	127-130	131-133	134-137	138-139	NQ	NQ
IV. Adults									
Genetic	161-162	163-164	NQ	NQ	NQ	172	NQ	175	NQ
Specialty	161-162	165-166	NQ	NQ	NQ	172	NQ	176	NQ
Pre-Sym	161-162	167-169	170-171	NQ	NQ	172	173-175	NQ	NQ
V. Public Health									
Newborn	201	206-208	215-222	228-233	235	NQ	238	239	NQ
Sickle Cell	202	209-211	223-225	234	236	NQ	238	239	NQ
Genetic	203	212-214	226-227	NQ	237	NQ	238	239	NQ
Registries	204-205	NQ	NQ	NQ	NQ	NQ	238	239	240-245

KEY: NQ – NO QUESTIONS